HK J Paediatr (new series) 2022;**27**:46-84

Proceedings of Congress

Joint Annual Scientific Meeting 2021

The Hong Kong Paediatric Society, Hong Kong College of Paediatricians, Hong Kong Paediatric Nurses Association, and Hong Kong College of Paediatric Nursing

30 October, 2021

Oral Presentation

Development, Implementation, and Evaluation of a Clinical Practice Guideline for Care of Preterm Infants Receiving Non-Invasive Ventilation: A Before and After Study

SY Chan,[1] J Chau,[2] KL Siu,[1] B But,[1] YM Yeung[1]

[1]Department of Paediatrics, Queen Elizabeth Hospital; [2]The Nethersole School of Nursing, The Chinese University of Hong Kong, Hong Kong SAR

Background: Nowadays, noninvasive ventilation is the mainstay of the ventilation strategy in the neonatal intensive care units (NICUs) and most of infants, especially preterm infants, having respiratory problems, are provided noninvasive ventilation (NIV) upon their demands. Nevertheless, complication of NIV device-related pressure injury was common, the incidence of nasal injury ranged from 20% to 60%. Limited studies were found evaluating the nursing care of preterm infants receiving NIV.

Aims: This study aimed to develop an evidence-based clinical practice guideline for preterm infants receiving NIV, implement the guideline in a NICU of a regional hospital, and evaluate infant outcomes including comfort, incidence of NIV device-related pressure injury. Besides, improvement on nurse's knowledge and practice for caring infants under NIV were assessed.

Study Design and Methods: The Iowa Model-Revised was adopted as the theoretical framework to guide the study process. A multidisciplinary workgroup consists of eight stakeholders in NICU was formed for the process and acted as the champions for the new practice. A before and after study design was adopted and included the pre-implementation and post-implementation phases. An integrative review was conducted to identify relevant studies from eight electronic databases before the study. All eligible studies were appraised using the Johns Hopkins University's evidence appraisal tool. Neonatal Pain, Agitation and Sedation Scale (N-PASS) for pain assessment and two self-developed NIV care bundle knowledge test and audit tool were used for the study.

Results: Due to the COVID-19 pandemic in 2020, the study was extended for a month and ended in January 2021. A total of 74 infants in Pre-implementation phase (before group) and 67 infants in Post-implementation phase (after group) were recruited. Logistic regression model was used to compare the incidence of pressure injury between groups after adjusted for all substantial covariates in the study. Infants in after group had an 84% decreased odds of acquiring pressure injury (adjusted OR=0.149, 95% CI 0.045-0.495, p=0.002). Infant's comfort level whilst receiving NIV was not determined in the study as the after group having a significantly lesser mean time ($p<0.001$) in calm state but lower N-PASS score.

Regarding nurse participants, 71 nurses received the training programme on NIV care bundle, and overall nurses' knowledge level improved immediately (adjusted $p<0.001$) and at 12 weeks after the programme. Three audits were conducted to evaluate nurses' practice, nurses' compliance rate to the care bundle significantly improved at 12 ($p<0.001$) and 24 weeks ($p<0.001$) in comparison with baseline compliance rate in the pre-implementation phase. However, nurses' knowledge retention at 12-week and compliance rate at 24-week after the training programme declined.

Conclusion: The evidence-based clinical practice guideline aims to promote comfort and prevent injury in infants receiving NIV, and outcomes of the infants depend on vigilant nursing care and compliance to this clinical practice guideline. Declining of nurse's knowledge level and practice compliance found in the study indicates the needs of continuous education and audit on the practice to sustain the service quality and patient's safety.

Surgical Intervention Reduces Long-Term Heart Rate Variability During Sleep in Children with Moderate-To-Severe OSA: A Secondary Analysis of an RCT

KTW Cheung, NHY Chow, KCC Chan, AM Li, CT Au

Department of Paediatrics, Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR

Background: Childhood obstructive sleep apnoea (OSA) is associated with heart rate variability (HRV) but there is no randomised controlled trial to evaluate the effect of OSA treatment on HRV in children.

Purpose: To investigate the effect of surgical intervention on HRV in children with OSA.

Methods: A prospective randomised controlled study was performed in non-obese pre-pubertal children aged 6 to 11 years with polysomnography (PSG) confirmed moderate-to-severe OSA. They were assigned randomly to early surgical intervention (ES) or watchful waiting (WW). The surgical intervention consisting of tonsillectomy with or without adenoidectomy and turbinate reduction was

carried out within 4-6 weeks after randomisation. Both groups underwent PSG with ECG monitoring at baseline and 9-month follow-up. Standard time- and frequency-domain variables of HRV were calculated.

Results: A total of 155 participants were randomised. Data of 52 subjects from the ES and 39 from the WW group was available for this secondary analysis. A significantly greater reduction in obstructive apnoea hypopnoea index (-10.6/h±1.5 c.f. -1.2/h±1.7, $p<0.001$) were seen in the ES group. For HRV parameters, notably greater reductions in the power of very-low frequency (-52.5 ms2±16.9 c.f. +9.4 ms2±19.5, $p=0.010$) components during stage N3, and SD of the mean of RR intervals in 5-min segments (SDANN) (-2.1 ms±2.0 c.f. +3.7 ms2±2.3, $p=0.026$) during stage REM were observed in the ES group, when compared to the WW group.

Conclusion: Surgical intervention could lead to a reduction in long-term fluctuations in the R-R intervals in children with OSA. It suggests that surgical treatment may reduce the activation of the sympathetic nervous system during sleep.

Acute Kidney Injury in Relation to Nephrotoxic Medication Use Among Critically Ill Children in the Paediatric Intensive Care Unit (PICU)

VPY **Chan**,[1] WF **Hui**,[2] VKW **Lok**,[1] HK **Tse**,[1] C **Wong**,[1] SM **Wong**,[1] KL **Hon**,[2] MH **Poon**[1]

[1]Department of Pharmacy; [2]Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Background: Children in the Paediatric Intensive Care Unit (PICU) are vulnerable to acute kidney injury (AKI) and due to the complex nature of the disease, they are often exposed to multiple medications which may individually or in combination have the potential to cause addition risk for renal injury.

Purpose: We presented the result of the interim analysis of an ongoing prospective cohort study on the potential association between nephrotoxic medications and the risk of developing AKI in critically ill children admitted to PICU of Hong Kong Children's hospital (HKCH).

Method: Patients were included if they were aged >1 month to ≤18 years of age and admitted to PICU of HKCH since 6/2020. Patients were excluded if they had pre-existing chronic kidney disease or impaired renal function for ≥3 months prior to PICU admission or admitted for post-renal transplant. The medication records from 14 days prior to PICU admission to PICU discharge would be retrieved and reviewed by an independent pharmacist to determine the number and doses of nephrotoxic medications exposure in relation to the development of AKI. The results of the initial four months of data would be presented.

Findings: A total of 62 patients with 63 admissions fulfilling the study criteria were identified. The overall incidence of AKI during PICU stay was 48.4% (Stage 1: 20.3%; Stage 2: 12.5%; Stage 3: 15.6%). 76.6% of the patients were exposed to one or more of 43 nephrotoxic medications. Altogether 17 (48.5%) of patients with AKI received nephrotoxic medications before development of AKI. The median number of nephrotoxic medication exposure was 1.5 (1, 3). The total medication doses received was 9.5 (1.0, 31.8) doses. Children with AKI were associated with significantly higher nephrotoxic medication exposure during the PICU stay. Patients with AKI received a significantly higher total number of nephrotoxic medications (3 vs 1 medication, $p<0.01$) and a higher total dose of nephrotoxic medications (21.5 vs 1.0 doses, $p<0.01$) than those without AKI. Furosemide, vancomycin and co-trimoxazole were the three nephrotoxic medications with the highest total administered doses. Cyclosporine A, foscarnet and ganciclovir were only given to those who had developed AKI.

Conclusion: AKI was commonly encountered among critically ill children in PICU. Critically ill children received a higher number and doses of nephrotoxic medications are at a higher risk of developing AKI. These patients should be monitored frequently and judicious use of nephrotoxic medications should be encouraged.

Unrevealing Parental Mosaicism: The Hidden Answer to the Recurrence of Apparent *De Novo* Mutations

M **Lee**, A **Lui**, CCY **Mak**, PHL **Doog**, JFT **Chau**, MHY **Tsang**, MMC **Chui**, CCY **Chung**, AKY **Kwong**, BHY **Chung**

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

Background and Purpose: Mosaicism refers to the co-existence of two or more genetically distinct cell populations in an individual from a single fertilised egg. Previous literatures estimated the percentage of parental mosaicism in rare diseases to be 4-8% (Myers et. al., 2018

and Breuss et. al., 2020), with a much lower percentage locally at less than 1% (unpublished data). In this study, we aim to investigate hidden parental mosaicism in local rare diseases families.

Methods: Parents whose children had previously diagnosed with neurodevelopmental disorders with an apparent *de novo* variant identified through trio sequencing are being recruited (N=15). Blood, buccal swab, and semen sample are being used to detect potential parental mosaicism by sanger sequencing and droplet digital PCR, complemented with the blocker amplification method when possible.

Findings: We report here 2 positive results that demonstrated asymptomatic parental mosaicism with 2 mechanisms: (1) Somatic mosaic mutation is observed in the father of two affected fetus with CHARGE syndrome. The splice site variant CHD7:c.7164+1G>A was found in both affected fetus and in the semen of the father but not detected in paternal lymphocytes. Such mutation might have arisen during spermatogenesis due to paternal age effect, given the age of father at conception is already 36 years old. (2) Gonosomal mosaic mutation in FLNC: c.4916G>A (p.Cys1639Tyr) is observed in the semen, buccal and lymphocytes of the father of two children with restrictive cardiomyopathy, demonstrating that the mutation had arisen from early embryogenesis.

Conclusions: Obtaining the right sample type and implementing sensitive detection method to identify mosaicism is currently limited but of great importance. Carrying a hidden mutation in the germ cells will have an impact on reproductive risk for next pregnancies, which could have a recurrence rate of as high as >50% (Shu et. al., 2021).

Acknowledgement: This study is supported by the Society for the Relief of Disabled Children and the Seed Funding for Basic Research (201910159064) from the University of Hong Kong.

Incidence and Outcome of Electrolytes Disturbances Among Critically Ill Children

WF **Hui**, KL **Hon**

Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Electrolyte disturbances are common yet overlooked conditions in paediatric critical care. The local incidence and outcome of electrolytes disturbances among critically ill children are largely unknown.

Purpose: We described the epidemiology of electrolytes disturbances among children admitted to the paediatric intensive care unit (PICU) of the Hong Kong Children's Hospital. This is the interim analysis of the prospective cohort study (E-AKI-DRUG) conducted in our PICU.

Methods: All children aged 1 month to 18 years old would be enrolled. Exclusion criteria included those with pre-existing chronic kidney disease, impaired renal function for ≥3 months, immediate post-renal transplant and short PICU stay of ≤1 days. AKI would be defined using the KDIGO criteria. The serum electrolytes profiles on sodium, potassium, calcium, phosphate and magnesium were reviewed. Appropriate urinary investigations were performed to look for tubular dysfunction among those with electrolytes disturbances. The data of initial four months would be presented.

Findings: We identified 63 episodes of admission for analysis. 59% were male and the median (interquartile range) age was 6.1 (6.6) years old. 49.2% of patients had an oncological diagnosis and 9.5% were recipient of bone marrow transplantation. The median number of types of electrolytes disturbances was 4 (3) types. Hypophosphatemia (85.5%), hypocalcaemia (77.4%) and hypokalaemia (61.3%) were the three types of electrolyte disorders with highest incidences. Children with AKI ($p<0.001$), requiring inotropic support ($p<0.01$) and mechanical ventilation ($p<0.01$) had higher number of electrolytes disorders. Tubular dysfunction was common among children with electrolytes disturbances. The proportions of children with urinary wasting of potassium (25%), phosphate (50%) and magnesium (87.5%) were high among those with hypokalaemia, hypophosphataemia and hypomagnesaemia. Abnormal urinary beta-2-microglobulin (median level of 0.9 [5] μg/ml) occurred in 64.7% of children with ≥2 types of electrolytes disturbances. Tubular dysfunction may occur independent of AKI as various urinary indices showed no significant difference between those with and without AKI. The number of electrolytes disturbances were associated with increased duration of ventilation ($p=0.011$) and PICU length of stay ($p<0.001$) and higher risk of PICU mortality (relative risk 4.3 [95% CI 1.4, 12.7]).

Conclusion: Electrolytes disturbances were common among critically ill children and may contribute to PICU morbidity and even mortality. Proximal tubular dysfunction was associated with multiple electrolytes disturbances.

Relationship Between Eczema Severity and Skin Microbial Biodiversity and Compositions in Hong Kong Infants

TF Leung,[1,2] YH Chen,[1] KCC Chan,[1] ASY Leung,[1] ZG Chen,[3] JWK Yau,[1] PKS Chan,[3] WH Tam[4]

Departments of [1]Paediatrics; [3]Microbiology and [4]Obstetrics and Gynaecology, Prince of Wales Hospital; and [2]Hong Kong Hub of Paediatric Excellence, The Chinese University of Hong Kong, Hong Kong SAR

Background: Eczema is the commonest chronic skin disease in children. Skin microbiome modulates the susceptibility and severity of childhood eczema, but there is limited data on its role for eczema severity in infants.

Objective: To characterise early-life skin microbial profiles and its effect on infantile eczema.

Methods: This birth cohort followed 120 Chinese infants at 1, 6, 12 and 24 months. Eczema diagnosis was made according to Hanifin and Rajka criteria, and severity was assessed using SCORing Atopic Dermatitis (SCORAD). Patients with objective SCORAD ≥15 were categorised as having moderate-to-severe eczema. Serial skin swabs taken at left antecubital fossa were subjected to 16S rRNA sequencing, and differentially abundant taxa for eczema phenotypes were analysed by Analysis of Compositions of Microbiomes with Bias Correction.

Results: Twenty-nine, 29, 7 and 3 subjects in this cohort had moderate-to-severe eczema at 6, 12 and 24 months respectively. Compared with mild eczema at 24 months, patients with moderate-to-severe eczema had lower alpha diversity of skin microbiome at 6 months as indicated by Shannon (P=0.03) and Simpson (P=0.03) biodiversity indices. The relative abundances of Janibacter (adjusted P<0.001) and Acinetobacter (adjusted P<0.001) at 1, 6 and 12 months were consistently lower in patients who had moderate-to-severe eczema than those with mild eczema at 12 and 24 months old.

Conclusions: Skin microbial biodiversity and compositions during infancy may predict the presence of moderate-to-severe eczema by 24 months in Chinese toddlers. (Funded by Health and Medical Research Fund [reference no. 06170466])

Identification of Common Variants in Vitamin D Binding Protein (GC) and Vitamin D Receptors (VDR) in Affecting Serum 25(OH)D Level in Han Chinese Healthy Infants and Toddlers

HW Tsang, K Tung, BNK Chan, SY Wong, P Ip

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

Background: Infancy represents an important period of development and vitamin D plays an essential role in it. Hypovitaminosis D is associated with long-term health issues and diseases development in children and infants. Recent findings in Vitamin D genetics illustrated an important role in determining serum 25(OH)D concentration, with genes controlling vitamin D metabolic pathway. Specifically, GC gene and VDR gene, encoding the vitamin D binding protein and vitamin D receptor are both involved in vitamin D metabolism and found to be associated with serum 25(OH)D. While associated key environmental factors have been extensively identified in children, adolescent and adults, the effects of genetic variation in relation to the serum vitamin D in Han Chinese infants warrant further exploration.

Objective: This study aimed to examine the effects of genetic variability in vitamin D binding proteins (GC) and vitamin D receptor (VDR) on the serum 25(OH)D levels among Chinese infants. A local set of hypovitaminosis D risk haplotype will be constructed to lay down the genetic basis specifically for the infants.

Method: Stratified random sampling was adopted to recruit infants and toddlers aged 2 to 24 months in the period of 1 June 2019 to March 2021. DNA was extracted from the collected whole blood samples and genotyping was performed by allelic discrimination and validated by Sanger Sequencing. Serum 25(OH)D was measured by Liquid Chromatography Tandem Mass Spectrometry (LC/MS-MS) and the measurement accuracy passed the proficiency testing of Vitamin D External Quality Assessment Scheme (DEQAS) by Endocrine Laboratory, London, UK.

Results: A total of four single nucleotide polymorphisms were genotyped among 243 infants. GCrs7041 and rs2282679 were significantly associated with serum 25(OH) D levels in Hong Kong infants, in which GCrs7041T and GCrs2282679C were identified as risk alleles. Further analysis on VDBP haplotype (rs7041-rs4588) revealed that the VDBP protein isoform GC1F was significantly associated with lowered 25(OH)D levels in the combined haplotype manner and GC1S demonstrated with the highest

amount of infant serum 25(OH)D. Carriers of GC1F-rs2228570C demonstrated a common negative association in serum 25(OH)D and identified as an interactive risk haplotype.

Conclusion: Genetic factor was identified as a key determinant of infant vitamin D level in Hong Kong infant. The risk rs7041 allele and corresponding VDBP isoforms contribute to a higher risk of vitamin D deficiency in infants. A potential interaction between VDBP haplotype and rs2228570 was found. Specifically, the effects of GC1F haplotype on serum 25(OH)D levels were found to be more dominant among carriers of rs2228570A. The risk haplotype VDBP GC1F-VDR2228570A was identified. This preliminary study provided infant-specific vitamin D genetics data in Han Chinese and laid down the foundation for post-natal screening in general public, preventing the risk of the genetic factors leading to vitamin D deficiency or insufficiency.

Tetraspanin CD9 Drives Immune Evasion and Disease Progression in Paediatric Acute Myeloid Leukaemia

Y Xu, KYY Chan, SP Fok, TK Man, CK Li, KT Leung

Department of Paediatrics, The Chinese University of Hong Kong, Hong Kong SAR

Background/Purpose: We recently reported the importance of CD9 in paediatric ALL but its role in AML remains unknown. Here we pursued to characterise its prognostic significance, elucidate its function, and identify its role in immunosurveillance.

Methods: Patients were stratified based on CD9 status for comparison of long-term survival. Functional impact of CD9 was measured by competition and colony formation assays. Influence of CD9 on leukaemia progression was evaluated in xenograft models, coupled with single-cell transcriptomics. Immunoregulatory role of CD9 was identified in an immune-reconstituted mouse model.

Findings: CD9 expression of AML patients was significantly lower than normal BM donors. Among 81 AML cases, blasts of 32 patients (39.5%) were CD9+. The 5-year RFS rate of CD9- patients was significantly lower than CD9+ patients. Overexpression of CD9 reduced proliferation and clonogenicity of AML cell lines. NOD/SCID mice receiving CD9+ AML exhibited a drastic reduction of leukaemic load. Mechanistically, CD9 promoted basal and cytokine-induced MHC I/II expression through the JAK-STAT axis. Importantly, CD9 could enhance BM infiltration of cytotoxic T cells and mount an effective immunity against AML in humanised NSG mice.

Conclusions: Our data established CD9 as a novel immune regulator in paediatric AML, and inspired a new treatment strategy for this rare but aggressive malignancy.

Impact of Personalised Survivorship Care Plan on Improving Awareness of Health Risks Among Survivors of Childhood Cancer: A Preliminary Analysis

CTJ Ma,[1] LS Yang,[1] HWJ Chou,[1] YTA Tung,[1] YWB Lai,[1] VWY Lee,[2] NCY Yeung,[3] SLK Lee,[4] AWK Leung,[4,5] CK Li,[4,5,6] YT Cheung[1]

[1]School of Pharmacy, CUHK; [2]Centre for Learning and Enhancement and Research, CUHK; [3]JC School of Public Health and Primary Care, CUHK; [4]Department of Paediatrics & Adolescent Medicine, HKCH; [5]Department of Paediatrics, CUHK; [6]Hong Kong Hub of Paediatric Excellence, CUHK, Hong Kong SAR

Introduction: We have previously reported that long-term survivors of childhood cancer in Hong Kong demonstrated low awareness in treatment-related late effects. There is emerging evidence in the Western literature that demonstrated improved health literacy in this population after the provision of a survivorship care plan.

Purpose: To evaluate the effectiveness of a personalised education program on improving survivors' knowledge of cancer-related health risks.

Methods: We recruited survivors (or caregivers) diagnosed with cancer before 18 years old and were ≥2 years post-treatment. Upon recruitment (T0), we assessed the awareness of their own cancer diagnosis, treatment history, and treatment-related health risks using a structured questionnaire. We then provided a personalised treatment summary and counselling on the potential therapy-related late effects as per the Children's Oncology Group guidelines. The participants' cancer-related knowledge was assessed again at 1-month post-intervention (T1). The Wilcoxon signed-rank test was conducted to evaluate changes in cancer-related knowledge scores from T0 to T1.

Results: The intervention was administered to 185 survivors (67.4% hematological cancer; current age 18.6 [SD=6.6] years old; range: 4 to 38 years) who were 9.0 [SD=5.0] years post-treatment. The current analysis included 82 participants (44.8%) who completed post-

intervention assessment (67.1% haematological cancer; current age 17.9 [SD=6.1] years old; range: 4 to 34 years). From T0 to T1, there is significant improvement in participants' diagnosis awareness score (72.0 [SD=27.5] *vs* 89.4 [SD=20.2]; $P<0.0001$) and late effects awareness score (22.9 [SD=22.8] *vs* 60.4 [SD=28.2]; $P<0.0001$). More survivors could identify their treatment-related health risks at post-intervention (13% vs 69.5%; $P<0.01$). Most participants agreed that the program has improved their understanding of potential late effects (n=67, 81.7%).

Conclusion: The personalised education program shows preliminary effectiveness in improving cancer-related knowledge among local survivors of childhood cancer. Future work includes evaluating whether the program may lead to better lifestyle and survivorship outcomes. (Funded by HMRF/RF Ref 03170047)

AI-Driven Delineation of Distinct Phenotypes Associated with N-Terminal Truncations of the *MN1* Gene - Beyond a New Syndrome Discovery from Hong Kong

CCY Mak,[1] TC Hsieh,[2] MMC Chui,[1] M Lee,[1] *MN1* International Study Group, PM Krawitz,[2] B Chung[1]

[1]Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR; [2]Institute of Genomic Statistics and Bioinformatics, University of Bonn, Bonn, Germany

Background: *MN1* C-Terminal Truncation (MCTT) syndrome is a rare autosomal dominant disorder characterised by intellectual disability, mid-face hypoplasia, severe expressive speech delay, and an atypical form of rhombencephalosynapsis (Mak et al. Brain 2020 and GeneReviews®). The *MN1* gene is comprised of only two exons. Individuals with distinct features of MCTT harbour truncating variants at the C-terminal (within exon 2 or the last 55bp of exon 1), which are predicted to escape nonsense-mediated decay (NMD). Individuals affected by N-terminal (MNTT) truncations (predicted to induce NMD) have milder developmental phenotypes than MCTT patients.

Methods: Since our discovery of the syndrome, an expanded clinical case series was recruited to review 45 subjects (mean age 12.9, range 2-44) from North America, Europe and Asia. We performed deep phenotyping on patients affected by MNTT (n=13) and MCTT (n=32) mutations both clinically and using AI-based facial recognition software GestaltMatcher (Hsieh et al.). GestaltMatcher trains deep convolutional neural networks on 22,619 frontal images with 299 different rare disorders to learn the facial features, and it further converts facial images into feature vectors to form a Clinical Face Phenotype Space. The facial syndromic similarities among the patients are quantified by cosine distance in this space.

Results: Delineation of phenotype both clinically and by GestaltMatcher identifies two distinct groups when comparing MNTT with MCTT. Clinically, patients with MNTT have unique facial features, a disproportionate abundance of cleft palate (33% vs 7%) and conductive hearing loss (82% vs 35%). Compared to the MCTT group where 33% of individuals with MCTT rely on non-verbal communication only, and the remaining expressing first words at the mean age of 4.03 (range 2-6.75 years). Speech delay is less severe in the MNTT group with mean age first words at 2 (range 1.3-3 years). The distinction of MNTT and MCTT is supported by GestaltMatcher where clustering of MNTT and MCTT facial gestalt is observed and delineates from other syndromes in an unsupervised manner. Using this approach, GestaltMatcher also helps identify atypical cases where the phenotype does not follow the predicted rule of NMD.

Conclusions: Truncating mutations can have a region-specific effect on phenotype. Supported by AI-based approaches, MNTT and MCTT are two distinct facially recognisable syndromes in the same gene and are distinct from other known syndromes.

Acknowledgement: This study is supported by the Society for the Relief of Disabled Children.

Enhanced Pain-Management for Children Undergoing Cleft-Palate Repair to Shorten the Length-of-Hospital-Stay

KY To,[1] WM Wong,[1] KM Liu,[1] CF Kwok,[1] YT Mok,[1] MWY Tsui,[2] VMY Yuen,[2] PMY Tang,[1] NSY Chao,[1] MWY Leung[1]

[1]Department of Surgery; [2]Department of Anaesthesia, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Children with cleft-palate undergo primary repair in their first year of life. Postoperatively children stay in-hospital until achieving full feeding with satisfactory wound. Optimal pain-control is a key element influencing feeding, thus length-of-hospital-stay (LOS).

Standardised enhanced pain-management program after palatoplasty was implemented as collaborative **E**nhanced-

Recovery-**A**fter-**S**urgery project, promoting early discharge postoperatively.

Purpose: To provide an interim-review of the 'ERAS-effectiveness' of this enhanced pain-management program after palatoplasty.

Methods: Setting-up standardised postop-medication protocol for analgesia (+ antibiotics)

- Post-op-Day(POD)0-1: regular IV-Paracetamol + PR-Diclofenac (+ IV-Esmeprazole)
- POD2+: regular PO-Paracetamol and Ibuprofen (+Famotidine). If unfeasible oral route, continue IV-Paracetamol + PR-Diclofenac
- Rescue analgesic for breakthrough pain: IM-/PO-Tramadol (+ Ondansetron)

All patients undergoing palatoplasty after program implementation were prospectively included. Parents were given questionnaires to chart pain-score, vomiting, feeding-tolerance and need of extra analgesics on POD0-3.

Findings: Fourteen patients recruited, 14 questionnaires received.

64% reported minimal/mild pain only on POD1. 78% tolerated oral-fluids on POD1. 78% discharged on POD2/3. Nil reported significant breakthrough pain. All parents expressed satisfaction towards pain-control and early discharge.

57% reported moderate-to-severe pain with no rescue medication given. Hypotheses:

- Parents unaware of available rescue medication
- Reluctant/afraid about extra analgesics
- Nurses could not identify correct timing for rescue analgesic for breakthrough pain
- Promulgation/education on using rescue medication is important, to both parents and nurses

Conclusions: This project aims to enhance recovery after palatoplasty in children. Preliminary review suggests the pain-protocol shortens LOS by expediting adequate feeding via optimal pain-control. Future improvement measures and further studies to substantiate findings are considered.

Validation of the Sonographic Measurement of Lateral Parapharyngeal Wall Thickness in Children

HM **Yuen**,[1] CYA **Lai**,[1] KH **Liu**,[2] MC **Lee**,[2] KCC **Chan**,[1] WCW **Chu**,[2] AM **Li**,[1] CT **Au**[1]

[1]Department of Paediatrics; [2]Department of Imaging and Interventional Radiology, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong SAR

Background: Lateral parapharyngeal wall (LPW) thickness is a potentially useful anatomic marker of childhood obstructive sleep apnoea (OSA). Measuring LPW thickness by ultrasonography (USG) is technically feasible but its validity has not been verified in children.

Purpose: To assess the intra- and inter-operator reliability of the sonographic measurement of LPW thickness in children and to validate the measurement against MRI measurement.

Methods: Prepubertal children aged 6-11 years with suspected OSA were recruited. Using USG, LPW thickness was measured as the distance between the internal carotid artery and the echogenic surface of the pharynx in an oblique coronal plane. The measurement was repeated on the same day by the same operator twice and by another operator. By MRI, oblique dimension of LPW was measured at the retropalatal level. Intraclass correlation coefficient (ICC) was used to examine the intra- and inter-operator reliability. The agreement between the LPW thickness measured by USG and MRI was assessed by ICC and Bland-Altman plot.

Findings: Thirty-two children (mean age: 8.83±1.58, 25 male) were recruited. The intra- and inter-operator reliability of the LPW thickness by USG were good (ICC = 0.91 and 0.84, respectively). The agreement between the USG-measured and MRI-measured LPW thickness was acceptable (ICC = 0.65). The Bland-Altman plot demonstrated a mean difference of 0.075 cm and a 95% limits of agreement from -1.10 to 1.25 cm.

Conclusions: The sonographic measurement of LPW thickness in children demonstrated good intra- and inter-rater reliability and acceptable agreement with MRI measurement. Ultrasonography is a valid method to assess LPW thickness in children.

This study was supported by the Direct Grant for Research from the Research Committee of the Chinese University of Hong Kong (Project no. 2020.073).

The Impact of Nusinersen Treatment on Musculoskeletal Progression in Patients with Spinal Muscular Atrophy

HHN **Ip**,[1] MKY **Yu**,[1] PK **Cheng**,[1] KYH **Kwan**,[2] NLW **So**,[3] E **Kuong**,[3] SHS **Chan**[1*]

[1]Department of Paediatrics and Adolescent Medicine, Queen Mary Hospital, The University of Hong Kong; [2]Department of Orthopaedics and Traumatology, Queen Mary Hospital, The University of Hong Kong; [3]Department of Orthopaedics and Traumatology, The Duchess of Kent Children's Hospital at Sandy Bay, Hong Kong SAR

*Corresponding author

Background: Spinal muscular atrophy (SMA) is a neuromuscular condition associated with multiple comorbidities. Scoliosis and hip instability are common due to progressive deterioration and imbalance in muscle strength.

Purpose: Nusinersen, an intra-thecal delivered medication to treat SMA, has documented effects on improving strength and motor function. However, there is no published literature regarding the long term effects of nusinersen on scoliosis progression and hip instability. Our study aims to evaluate musculoskeletal progression of SMA patients after nusinersen treatment.

Methods: Under the current IRB approved prospective study, detail demographic and clinical information is gathered to document clinical outcome of all SMA patients undergoing nusinersen treatment. We excluded those who received surgery for the hip and/or spine within the study period. For all patients, scoliosis X-ray series were taken before the starting of nusinersen treatment and at regular intervals every 6 to 9 months. Cobb's angle was measured and average rate of progression per year post nusinersen treatment was calculated. For hip instability assessment, baseline X-rays taken at pre-nusinersen screening were compared to the latest in-patient X-rays using Reimer's migration index (RMI). Clinically significant changes were defined as 1) if hip changes from reduced to subluxed or dislocated and from subluxed to dislocated, 2) if RMI was <0.5 and progressed to >0.5, or 3) if RMI was >0.5 and progressed to >0.8.

Findings: By June 2021, a total of 24 SMA patients have started on Nusinersen treatment in our institution. Sixteen patients had scoliosis in their latest X-rays and 1 was excluded due to a short follow-up period. The median age of patients was 8.0 years (range 2.2-18.0 years). The average follow-up period was 27 months (range 15-47 months). At baseline prior to nusinersen treatment, scoliosis was present in 5 out of 6 SMA type 1 patients, 4 out of 7 SMA type 2 patients, and 2 out of 2 SMA type 3 patients. At final follow-up, 3 out of 5 SMA type 1 patients experienced curve progression, whilst 2 patients remained stable; all 4 SMA type 2 patients had progressed; and both SMA type 3 patients had curve progression. The average rate of Cobb's angle progression for type I patients was 6.9 degrees/year, type II patients was 13.6 degrees/year, type III patients was 5.2 degrees/year. Eighteen patients were included for hip analysis. Those who did not have a hip X-ray for comparison after the loading doses of nusinersen and those who underwent surgery were excluded. The median age of patients was 7.9 years (range 2.1-26.7 years). The average follow-up period was 22 months (range 14-36 months). RMI progressed in 10 out 16 hips in SMA type I patients, 7 out of 16 hips in SMA type II patients, and 2 out of 4 hips in SMA type III patients. In hips with RMI progression, 4 were clinically significant in type I, 3 were clinically significant in type II, and none were clinically significant in type III.

Conclusion: SMA patients treated with nusinersen show significant musculoskeletal progression, particularly in the more severe, non-ambulatory group. SMA patients on nusinersen should be placed on closer monitoring for scoliosis progression and hip instability. More aggressive interventions should be considered. Large scale studies with longer follow-up periods are warranted to confirm the findings of this study.

Investigating the Health-Related Quality of Life of Rare Disease Patients in Hong Kong

YNC **Ng**,[1] CCY **Chung**,[1] WHS **Wong**,[1] BHY **Chung**[1,2,3]

[1]Department of Paediatrics and Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong; [2]Department of Paediatrics and Adolescent Medicine, Queen Mary Hospital; [3]Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Background: Rare disease (RD) affects less than one in 2,000 individuals, often causing life-long physical or intellectual disability. In Hong Kong (HK), approximately 470 types of RDs were identified, affecting one in 67 people. Nonetheless, the health-related quality of life (HRQoL) of RD population as a group was never reported in HK.

Purpose: To investigate the HRQoL of RD patients in HK.

Methods: EuroQol 5-Dimension (EQ-5D) was used to describe HRQoL. RD patients and caregivers were

recruited through Rare Disease Hong Kong, the largest RD organisation in HK, between March and October 2020. The five health dimensions assessed were usual activities, pain/discomfort, mobility, anxiety/depression, and self-care. Utility scores were generated with reference to the HK value set. Meta-analysis was conducted using a random-effect model for studies investigating the HRQoL of RD patients using EQ-5D.

Findings: Overall, 289 independent participants were recruited, covering 116 unique RDs. The mean age was 31.6 (S.D. 19.8), where 30.8% were patients ≤18 years. The mean utility score of patients was 0.52 (S.D. 0.36), with 10.4% reported negative scores indicating worse-than-death health states. Patients with rare neurologic diseases had significantly lower mean scores than other patients ($p<0.001$). Comparing to existing literatures, RD patients had significantly lower mean utility scores than the general population (0.92), patients with diabetes (0.87), hypertension (0.88), heart disease (0.88), and cancer (0.87) in HK. Caregivers of RD patients also reported significantly lower utility scores compared to the HK general population (0.80 vs 0.92). In the meta-analysis including 6 studies comprising 2395 patients, the pooled mean utility score was found to be 0.58 (95% CI 0.47-0.69, I^2 98.8%).

Conclusions: This is the first study in HK to illustrate the significant impact of RDs on HRQoL, which warrants exceptional care from policy makers and society.

Acknowledgement: We would like to thank the Society for the Relief of Disabled Children for their funding support, and all patients and care-givers who participated in this study.

Poster Presentation

The Establishment and Benefit Impact of Donor Milk Bank of Hong Kong University Shenzhen Hospital: 8 Months Experience

QS Zhang, T Su, CB Chow

Department of Pediatrics, University of Hong Kong-Shenzhen Hospital, Shenzhen, Guangdong, China

Background: Mother's own milk (MOM) is the best choice for preterm infants and sick neonates. When MOM is not available or not enough, donor milk can be an alternative.

Purpose: To integrate the benefit and experience in establishing a Human Donor Milk Bank (HMB) in the Hong-Kong University ShenZhen Hospital to promote nutrition status and exclusive breastfeeding rate.

Method: A multidisciplinary team was established to prepare and manage the HMB. To ensure the safety of human donor milk, we adopted guidelines from Human Milk Banking Association of North America (HMBANA) and operation standard issued by the dietitian society of China. Several critical control points were identified for close monitoring including donor recruitment and donor milk pasteurisation. All donor milk can be traced back for its donation and processing information, such as name of donor, donor's virology status, date of donation, mode of sterilisation and handling staff etc.

Findings: Total 256 mothers donated 13,2800 mls of donor milk since operation of the HMB. Twenty-eight infants with mean GA of 30+2 weeks (SD±40.15 days) and mean birth weight 1510.93g (SD±948.27g) in NICU received donor human milk during hospitalisation, 82.14% of whom were VLGA. The average length of stay was 38.54 days (SD±32.54 days). An increased in the exclusive breastfeeding rate upon discharge was observed before and after the operation of HMB from 13% to 40%.

Conclusion: Through establishment of human milk bank and maintain a safe and sustainable management system is an important means to promote breastfeeding.

Implementation of a Family-Centered, Multidisciplinary Clinic for Early Diagnosis of Neurodevelopmental Impairment and Cerebral Palsy of Critically Ill Preterm Neonates in Shenzhen, China

HB Huang,[1] MJ Watt,[1,2] M Hicks,[3] QS Zhang,[1] F Lin,[1] XQ Wan,[1] CB Chow,[1] PY Cheung,[1,3] MDAC Program[4]

[1]Department of Pediatrics and [4]Physical Medicine and Rehabilitation, University of Hong Kong-Shenzhen Hospital, Shenzhen, 518000 China; [2]Department of Pediatrics and Department of Physical Medicine and Rehabilitation, Glenrose Rehabilitation Hospital, University of Alberta, Edmonton, Canada; [3]Department of Pediatrics, University of Alberta, Edmonton, Canada

Aim: To compare the time to diagnosis of neurodevelopmental impairment (NDI) and cerebral palsy (CP) in preterm neonates (<29 weeks) at a Multidisciplinary Assessment and Care (MDAC) clinic with that of a conventional high-risk infant follow-up clinic.

Methods: All eligible surviving preterm neonates born at <29 weeks gestation at the University of Hong Kong-Shenzhen Hospital between January 2015 and December 2019 were followed up in conventional (2015-2017) and MDAC (2018-2020) clinics up to 2 years of corrected age with clinical demographic information collected in a prospective database. The MDAC team used standardised developmental assessments. The rates and timing of diagnosing NDI and CP in two epochs were compared.

Results: The rates of NDI and CP were not different in two epochs (NDI: 12(50%) vs. 12(41%); CP: 3(12%) vs. 2(7%) of 24 and 29 surviving infants assessed in conventional and MDAC clinics, respectively). Infants followed up in the MDAC clinic were diagnosed with NDI and CP earlier than those in the pre-MDAC epoch (6 vs. 14 months corrected age, respectively, P<0.05).

Conclusion: High-risk preterm neonates can be followed more effectively in a family-centered, child-friendly multidisciplinary clinic leading to an earlier diagnosis of NDI and CP. Early counselling and interventions could be implemented accordingly.

Use of Different Device in Handling Haemodialysis Catheter to Streamline the Procedure Time and Catheter Revision Rate

KM Chan, SC Chan, KY Ko, CH Chan, PW Chung, MC Chan, KN Tsim, MS Liu, PC Or, VWC Lau, SK Tang, EYH Chan, ALT Ma, WM Lai

Nephrology Centre, Department of Paediatrics, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Different from adult renal patients, cuff central venous haemocatheter (CVC) are majority used in children on chronic haemodialysis (HD) in Hong Kong. A bundle of strategies is used to minimise the rate of central line- associated blood stream infection (CLABSI), thrombosis and catheter malfunction. In the past, a sterile reusable renal set and blind-end device is used to capped the CVC with prescribed anti-coagulant. Since the year 2020, A needle-free capping device (Tego connector, TC) was introduced to the current practice. After each HD session, a sterile disposable dressing set and a TC was use to cap the CVC. The current study was to compare the past and new practice of cap device in a haemodialysis center so to improve the patients' comfort and the procedure cost.

Methodology: The study was conducted from 1/3/2020 till 28/2/2021. The patients' inclusion criteria were those patients with a cuff CVC and the exclusion criteria of the study were those patients' CVC are incompatible with TC and arterial- venous fistula. The catheter function such as any blockage, thrombosis formation and rate of CLABSI was reviewed every HD session by renal nurses and monthly medical review.

Result: Total 14 patients were recruited in the study period. Eight female and 6 males whose age range was 3 years - 31 years. The CLABSI was remaining zero between two devices. No clot formation inside the catheter lumen and connector dislodgement was not detected after using the TC. The catheter revision rate in patients using TC was lower from 1.0 to 0.71 per 1000 catheter days as compared with using the blind-end capped. The current study showed a better catheter survival rate than the McAfree & Seidal study (2010) in USA 0.78 per 1000 catheter days. The disinfection procedure using a sterile disposable dressing set at the end of the HD session was reduced from 15 minutes to 10 minutes in each patient. It implies that the paediatric patients no need to hold the same position in every HD session for more than 15 minutes. In fact, holding same position for was not easy especially the younger age patients. Total nursing time saved was 174 hours in the study period. Furthermore, there are 40% decrease the usage of sterile reusable renal set which replaced by sterile disposable dressing set. It implies to save the administrative cost to handle sterile set from Sterile Supply Unit (SSU), ward manual daily checking.

Conclusion: In the study, the change of practice using TC for HD paediatric patients could promote the patients' comfort to reduce the time of holding same position during the HD therapy and the cuff CVC survival rate. The change of practice also streamlines the procedure time and decrease the administrative cost in handling reusable set.

A COVID-Adapted Neonatal Resuscitation Workshop for Teaching Neonatal Resuscitation – A Regional Hospital's Experience

KL Siu,[1] NM Chan,[1] PY Leung,[1] SF Lee,[1] SY Chan,[1] SY Cheung,[1] SH Leung[2]

[1]Department of Paediatrics; [2]Multi-disciplinary Simulations and Skills Centre (MDSSC), Queen Elizabeth Hospital, Hospital Authority, Hong Kong SAR

Background: A good and effective neonatal resuscitation training (NRT) program is important for retaining skills and achieving good outcome. A stimulation based Neonatal Resuscitation Workshop (NRW) has been held quarterly for Paediatric, Obstetric and Emergency Department trainees since 2011. The NRW was suspended since July 20 due to COVID-19 pandemic. A web-based COVID-adapted NRW (CaNRW) was developed in January 21 to achieve the objectives of providing NRT and abiding infection control measures.

Methods: A CaNRW was developed with the Multi-Disciplinary Simulation and Skills Centre (MDSSC), Queen Elizabeth Hospital in January 21. It consists of a 1-hour lecture delivered in YouTube; a 1- hour video on skills and a birth related drill performed at MDSSC. All trainees participate it online except those involved in the drill. Trainees can join the drill and debriefing online simultaneously. Upon competition, they were asked to complete an evaluation.

Results: Total 113 trainees joined three CaNRW in 21. Trainees didn't report difficulties in viewing. Facilitators felt more comfortable in executing this CaNRW. Seventy-three evaluations were analysed. Trainees reported this CaNRW was useful with high level of satisfaction (Score: 4.41/5). Trainees strongly recommended it to their colleagues (Score: 8.53/10). Nurses and midwives were granted Continuous Nursing Education and PEM credits respectively.

Conclusions: This innovative web-based CaNRW provides a simple and structured model for NRT during COVID-19 pandemic. Trainees welcomed and enjoyed it. Thus, this CaNRW is worth promoting and should be made available to more trainees and departments. More studies should be performed to testify its clinical values.

Measuring Compliance to Ketogenic Diet Among Obese Children and Adolescents

K Sun,[1] LL Hui,[2] EAS Nelson[1]

[1]Department of Paediatrics, The Chinese University of Hong Kong; [2]Department of Applied Biology and Chemical Technology, The Hong Kong Polytechnic University, Hong Kong SAR

Background: Ketogenic diet, low in carbohydrate and rich in fat, have been used for weight loss but compliance is challenging among obese children and adolescents. Assessing compliance is important for evaluating its effectiveness.

Purpose: To review reported compliance to ketogenic diet among obese children and adolescents.

Methods: A systematic search for literature was performed using PubMed to identify intervention studies on the effect of ketogenic diets among obese children or adolescents. Additional hand search was performed. Full text studies in English with primary data on compliance to ketogenic diet (claimed ketogenic or very-low-energy diet) were included. Subjects' characteristics, dietary prescription, duration, use of pre-made food, frequency of dietitian counselling, instructional menus, family involvement, self-reported side-effects, measures of compliance, calculated compliance and retention rate were extracted.

Findings: Eight papers are included. Majority were carried out in the U.S. on 6-to-18-year-old subjects. Intervention periods ranged from 6 weeks to 6 months. Recommended carbohydrate content was 10-60 grams per day. Four studies also limited fat and/or protein consumption. Subject retention rate was 57-100%. Most had weekly or bi-weekly dietitian counselling. Side effects including constipation, nausea and headache were generally mild and infrequent. One study provided pre-made food which seems to increase compliance (62.5%). Studies mainly used food records to measure compliance with 1 using >5% weight loss and 4 monitoring urinary/blood ketones.

Conclusions: Assessing the compliance to ketogenic diet is important in clinical studies and yet complicated. More convenient assessment of ketosis is required for design and analysis of studies on ketogenic diet.

Proactive Intervention Program for Pregnant Ladies with Substance Abuse at Antenatal Period

DCH Wan,[1] MY Lui,[2] DFY Chan[1]

[1]Department of Paediatrics; [2]Department of Obstetrics and Gynaecology, Prince of Wales Hospital, Hong Kong SAR

Background: Multidisciplinary case conferences (MDCC) will be conducted for every newborn with positive urine result of illicit drugs since 2016. Moreover, court hearing for care or protection order will be arranged for those require out of home care. In 2018, our team implemented a new program of screening illicit drugs for all pregnant ladies with history of substance abuse during antenatal period. Urine screening for toxicology were arranged at the first visit and antenatal welfare plan meeting will be attended by paediatrician and social workers during one of the antenatal checkup visit for those with positive urine result. Explanation of harmful effect of drugs to fetus, appropriate detoxification programs and child care plan will be discussed.

Objectives:

1) To reduce the harmful exposure of illicit drug to the fetus,
2) For early engagement of detoxification program to the addicted pregnant ladies,
3) To reduce the burden in conducting MDCC and Court hearing,
4) To reduce the length of hospitalisation of the affected newborns and
5) To increase the rate of discharge of babies back to drug-free mothers.

Methodology: Under Comprehensive Child Development Service (CCDS), all pregnant ladies with history of substance abuse were recruited prospectively with written consent in 2018. The mother-child dyad would be followed up until the child at 24 months.

Result: In 2018, there were 41 pregnant ladies of mean age of 28.2 years old with history of substance abuse recruited. The mean age for the first intake of illicit drug was 17.9 years old. Twenty-six (63%), 19 (46%) and 16 (39%) of them reported using ketamine, cocaine and

amphetamine respectively. Twenty-six (63%) of them were using multiple drugs at the same instant. Antenatal urine screening for toxicology were conducted in 30 (73%) of them and 11 of them refused to provide urine sample or poor attendance in further antenatal visits. Thirteen of them (43.3%) with at least one urine sample tested positive of any kind of illicit drugs during antenatal period. Ten of them (76.9%) quitted drugs after engaged to the proactive program. Moreover, the hospital stay of the babies was shortened from in average of 29 days to 8 days for those newborns with positive to negative urine. Refusal for antenatal urine screening and/or defaulting antenatal visits were predictive for active drug abusing as reflected by the high rate of drug being detected in the newborn (54.5%). Developmental assessments were performed in 40 children (97.5%) at the age of 24 months. Ten (25%) of them were diagnosed to have developmental delay with no correlation to the antenatal drug status. Three out of 18 (18.7%) from the group of mother tested negative were delay. Incident of developmental delay is higher than normal population (10%). Four ladies in this cohort were persistently active in taking drugs, three of them were under the "refused" group. Five of the mothers relapsed in taking illicit drugs and 2 of their children suffered from developmental delay. Five of the mothers had another pregnancy within next 36 months, two of the newborns had positive urine result of illicit drug at birth.

Conclusion: Antenatal proactive screening for substance abuse in pregnant ladies is effective in reducing (1) the drug exposure to fetus, (2) number of MDCC and court hearing, (3) length of hospitalisation of newborns. Moreover, it also increased the rate of abusing pregnant ladies in quitting the drug dependent habit. This program also served to identify those ultra-high risk ladies in terms of their refusal in conducting the screening test and poor attendance to antenatal visits. Higher incident of developmental delay is noted, possible the effect of perinatal drug exposure or the weak parenting skill might be the origin. Further long study would be needed for identify the cause.

Immunosuppressive Therapies in Children with Biopsy-Proven IgA Vasculitis Nephritis: A Tertiary Centre Experience

MHL **Lee**, EYH **Chan**, ALT **Ma**, WM **Lai**

Paediatric Nephrology Centre, Hong Kong Children's Hospital, Hong Kong SAR

Background: IgA Vasculitis Nephritis (IgAVN), also referred as Henoch-Schönlein purpura nephritis, can lead to persistent proteinuria in children. The aim of this study is to determine the efficacy of immunosuppressive therapies in the management of IgAVN in children.

Methods: A retrospective review was conducted for all IgAVN patients under the care of the Paediatric Nephrology Centre in Hong Kong between January 2009 and December 2019. Patients with biopsy-proven IgAVN with persistent moderate or severe nephrotic-range proteinuria despite Renin-angiotensin-aldosterone system inhibitor (RAASi), including angiotensin-converting enzyme inhibitors (ACEIs) and angiotensin receptor blockers (ARBs), were included. Patient demographics, clinical and laboratory data, details of medical treatment and clinical outcomes were evaluated.

Results: Of the 177 Chinese children with IgAV, 42 children developed proteinuria. Twenty-one patients (16 boys) had persistent proteinuria despite the use of RAASi. Kidney biopsy confirmed IgAVN at a median age of 8.5 years. At baseline, 3 (14%), 14 (66%), 3 (14%) and 1 (5%) patient had moderate proteinuria, nephrotic-range proteinuria, nephrotic syndrome and nephritic-nephrotic syndrome with renal impairment, respectively. Cellular crescents were found in 76% of renal biopsies.

All patients received oral corticosteroid at a median time of 33 days since the onset kidney involvement of IgAV. While 7 children (33%) with severe disease received monthly intravenous cyclophosphamide as induction therapy, 12 patients (57%) and 2 patients (10%) received calcineurin inhibitors and azathioprine, respectively. The maintenance therapy consisted of corticosteroid and one additional immunosuppressive agent, including calcineurin inhibitors (n=16, 76%), azathioprine (n=4, 19%) and mycophenolate mofetil (n=1, 5%).

Over a median follow-up period of 6.0 years, 18 patients (86%) attained complete remission at a median of 139.5 days since the initiation of immunosuppression. Upon the most recent follow-up, all patients had normal kidney function and the median UPCR was 0.11 mg/mg (IQR 0.10-0.16).

Conclusion: Immunosuppressive therapies were associated with favourable renal outcomes in children with biopsy-proven IgAVN presented with persistent moderate or nephrotic range proteinuria despite RAASi.

Pulmonary Complications in Premature Infants Using Beractant or Poractant for Respiratory Distress Syndrome in A Local Neonatal Intensive Care Unit

KL Wong, KL Siu

Department of Paediatrics, Queen Elizabeth Hospital, Hong Kong SAR

Background: Beractants or poractants are the two types of surfactants utilised in treatment of respiratory distress syndrome (RDS) in premature infants. This was a retrospective cohort study comparing the outcomes between the two groups of surfactants.

Purpose: It compared the incidence of pulmonary complications and neonatal outcomes of premature infants receiving beractants or poractants.

Methods: Seventy-two patients in beractant group, and 54 patients in poractant group, were included. The primary outcome was to measure the incidence of air leak syndrome (ALS) and pulmonary haemorrhage. The secondary outcomes assessed mortality, pulmonary performance and outcomes. Logistic regressions were performed to identify independent risk factors for significant primary outcome.

Findings: There was significantly higher incidence of pulmonary haemorrhage in poractant group, measuring 2.8% in beractant group and 13% in poractant group (p=0.038). The difference in the incidence of ALS was not significant (p=0.883). Presence of coagulopathy was the only significant independent risk factor (OR=18.672, 95%CI [1.681-207.450], p=0.017). Patients in poractant group had longer mechanical ventilation duration (p=0.019), duration of oxygen supplement (p=0.037), length of stay in hospital (p=0.005), and higher percentage of patients fulfilling the criteria of BPD (p=0.014).

Conclusion: The study showed higher incidence of pulmonary haemorrhage in poractant group. Only coagulopathy, but not the type of surfactant, was identified as an independent risk factor for pulmonary hemorrhage incidence in multivariate analysis.

Fracture Burden in Paediatric End Stage Kidney Disease

CS Ho,[1,2] YH Chan,[1,2] E Yu,[3] HL Lee,[1] YL Tung,[4] LT Ma[1,2]

[1]Paediatric Nephrology Centre, Hong Kong Children's Hospital; [2]Department of Paediatrics and Adolescent Medicine, Princess Margaret Hospital; [3]Clinical Research Centre, Princess Margaret Hospital; [4]Endocrine Team, Department of Paediatrics, Hong Kong Children's Hospital, Hong Kong SAR

Background: Paediatric patients with chronic kidney disease are known to have an increased risk of fracture, however relevant data is limited.

Purpose: To determine the incidence of fracture and associated factors in children with end stage kidney disease (ESKD) receiving renal replacement therapy (RRT).

Methods: A retrospective review on all paediatric patients with ESKD at the tertiary Paediatric Nephrology Centre in Hong Kong. Children who presented before 18 years with active follow-ups for 12 or more months by November 2020 were included.

Results: RRT was initiated in 69 children (55% boys), with 21 (30.4%), 10 (14.5%) and 38 (55.1%) patients received peritoneal dialysis, haemodialysis and kidney transplant. 10 fracture episodes were observed in 7 patients (10.1%) at a mean duration of 7.8 years since RRT initiation, corresponding to a cumulative fracture incidence of 227.8 per 10000 patient year. This rate was 5-folds higher than the published data from our local general paediatric population (45 per 10,000 person-years; p=0.01).

Children who sustained fractures were significantly younger at the time of RRT initiation (3.5 vs 10.4 years; p=0.02) with a longer time on dialysis (12 vs 2.7 years; p<0.001). Other associated factors included metabolic bone disease, difficulty in walking, radiological evidence of renal osteodystrophy, parathyroid hyperplasia/adenoma, a higher parathyroid hormone level and more use of cinacalcet.

Conclusions: Children with ESKD receiving RRT have a higher risk of fracture. Longer duration of dialysis and a higher parathyroid hormone level were potential modifiable factors associated with fractures.

Five Years Data on Department of Health Child Assessment Service (CAS) Preschool Children at Risk of Dyslexia and Their Comorbidities

KY LAI, CCC LAM, ML TANG

Department of Health, Child Assessment Service (CAS), Hong Kong SAR

Background: Dyslexia is known to be associated with a number of comorbidities, including developmental language disorder (DLD), attention-deficit/hyperactivity disorder (ADHD), and developmental coordination disorder (DCD). Previously, most reading at risk cases could not be identified before school age. With the development of new assessment tools including the Hong Kong Dyslexia Early Screening Scale (HKDESS) (2016) and Hong Kong Comprehensive Assessment Scales for Preschool Children (HKCAS-P) (2014), the number of preschool children shown to be at risk of dyslexia increased dramatically.

Purpose: This study reviewed CAS cases diagnosed in the recent 5 years as being "at risk of dyslexia" together with their comorbidities, with the purpose of enhancing vigilance to coexisting developmental problems for early management.

Methods: A search was performed on the CAS database. Data on children coded as "at risk of dyslexia" were retrieved from June 2015 to May 2020. Comorbidities of ADHD, DLD, and DCD diagnosed during preschool or at subsequent assessments during school age were searched and analysed.

Findings: A total of 5689 children were diagnosed as "at risk of dyslexia" for the 5-year period. Their ages at diagnosis ranged from 4 years 6 months to 6 years 2 months. The prevalence of comorbidities in these children were as follows: (i) ADHD 41.1% (n=2338), (ii) DLD 46.7% (n=2659), (iii) DCD 23.1% (n=1312).

Conclusions: As early as pre-school age, children at risk of dyslexia showed high prevalence of developmental comorbidities. The data provides an overall picture of this disease pattern in Hong Kong for future clinical and educational services planning.

Improved Neonatal Outcomes by Multidisciplinary Simulation - A Contemporary Practice in the Demonstration Area of China

C XU,[1] PY CHEUNG,[1,2,3] Y XUE,[1] CB CHOW,[1] Q XIE,[4] C DONG,[1] Q ZHANG[1]

[1]NICU, The University of Hong Kong-Shenzhen Hospital, Shenzhen, China; [2]Centre for the Studies of Asphyxia and Resuscitation, Neonatal Research Unit, Royal Alexandra Hospital and [3]NICU, University of Alberta, Edmonton, Canada; [4]Department of Obstetrics, The University of Hong Kong-Shenzhen Hospital, Shenzhen, China

Aim: To investigate the impact of collaborative in-situ neonatal resuscitation simulation in reducing the incidence of neonatal asphyxia and related morbidity.

Methods: Neonatal in-situ resuscitation simulation training has been conducted by neonatal and obstetrical collaboration once per week in the University of Hong Kong – Shenzhen Hospital (HKU-SZH) since 2019. Each simulation was led by two instructors, accomplished by three health care providers from obstetrics and neonatal intensive care unit, followed by participatory feedback by the providers and several observers. The incidence of neonatal asphyxia, severe asphyxia, hypoxic-ischemic encephalopathy (HIE) and meconium aspiration syndrome (MAS) before (2017-2018) and after (2019-2020) the regular simulation was analysed.

Results: There were 82 simulation cases including resuscitation of preterm neonates with different gestational age, fetal distress, meconium-stained amniotic fluid, congenital heart disease, etc. After multidisciplinary in-situ simulation, the incidence of neonatal asphyxia decreased from 8.4‰ to 6.4‰ (P=0.045). The incidence of HIE and MAS dropped from 1.0‰ to 0.1‰ (P=0.003), from 1.9‰ to 0.87‰ (P=0.014), respectively.

Conclusion: Weekly collaborative resuscitation simulation improves acute neonatal outcomes, with decreased rate of neonatal asphyxia, hypoxia ischaemic encephalopathy and meconium aspiration syndrome. Implementation of regular resuscitation training is feasible and may improve the quality of neonatal resuscitation with better neonatal outcomes.

Discuss and Share Experiences in Implementing FCC Models in University of Hong Kong – Shenzhen Hospital (HKU-SZH)

Y Xue,[1] YZ Chen,[1] QS Zhang,[1] CB Chow,[1] GP Lee,[1] PY Cheung[1,2]

[1]Department of Pediatrics, The University of Hong Kong-Shenzhen Hospital, Shenzhen, 518000 China; [2]Department of Pediatrics, University of Alberta, Edmonton, Canada

Aim: To discuss and share experiences in implementing Family centered care (FCC) models in Hong Kong - Shenzhen Hospital (HKU-SZH).

Methods: At present, FCC has not been well implemented in China and most NICU adopt closed management. Since December 2013, our department has introduced the FCC concept and implemented 24-hour opening ward, providing family members with a series of services including ward rounds, progressive health education, palliative care, breast-feeding, cold chain support of breast milk, breast milk bank, family visit, follow-up after discharge, and mutual support group for family members. The above services were maintained during the COVID-19 period.

Results: 10,615 newborns were treated from December 2013 to December 2020, the minimum gestational age of preterm infants treated successfully was 22^{+6} weeks and the minimum weight was 500g. The patient satisfaction rate in 2020 was 99.9%, and the average breastfeeding rate in 2020 was 84% while 83.4% of full-term infants and 90.2% of premature infants were breast-fed. (A multi-center survey of 974 cases of nutrition-related status of preterm infants in NICU in China reported that 13.6% of preterm infants were breast-fed during 2005-2006) The nosocomial infection rate from 2014 to 2020 was controlled between 0.41% and 2.02%. Literature showed that the incidence rate of nosocomial infection in neonatal care units of 17 grade A general hospitals in China from 2013 to 2014 was 3.35%.

Conclusion: An open NICU based on the FCC concept is feasible and contributes to increased breastfeeding rates and patient satisfaction without increasing the incidence of nosocomial infections.

Hypoxic Obstructive Sleep Apnoea in Children: A Subtype Associated with Higher Cardiovascular Risk

SYS Cheng, KCC Chan, AM Li, CT Au

Department of Paediatrics, Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR

Background: Childhood obstructive sleep apnoea (OSA) is a heterogeneous disease with variable clinical and polysomnographic manifestations that may contribute to the individual differences in cardiovascular outcomes even in patients with a similar level of conventional severity indexes.

Purpose: To investigate the differences in blood pressure (BP) outcomes between children with hypoxic and non-hypoxic OSA.

Methods: This retrospective study reviewed the records of OSA patients aged 5 to 14 years who had undergone both overnight sleep study and 24-h ambulatory blood pressure monitoring in Prince of Wales Hospital from July 2014 to December 2020. A patient was defined as having hypoxic OSA when >50% of the respiratory events were associated with a drop of ≥3% in oxygen saturation level. Patients with mild (obstructive apnoea hypopnoea index (OAHI) 1-5/h) and moderate-to-severe (OAHI ≥5/h) OSA were analysed separately.

Results: Respectively 99 (45 with hypoxic OSA) and 44 (23 with hypoxic OSA) patients with mild and moderate-to-severe OSA were included in this analysis. No significant differences in daytime and nighttime BP were observed between hypoxic and non-hypoxic OSA groups in both mild and moderate-to-severe subgroups. However, patients with hypoxic moderate-to-severe OSA had significantly less nocturnal dipping of both systolic BP (7.7%±4.7 c.f. 10.7%±4.1, p=0.035) and mean arterial pressure (8.2%±5.9 c.f. 11.9%±5.7, p=0.047) than the non-hypoxic counterparts despite having similar age, gender distribution, body mass index z score and OAHI.

Conclusion: Children with moderate-to-severe OSA and respiratory events that were predominately associated with oxygen desaturations had less nocturnal BP dipping, suggesting hypoxia might have an independent effect on cardiovascular risk.

The Demographics and Long-Term Outcomes of Paediatric Kidney Replacement Therapy in Hong Kong

EYH Chan,[1] WHS Wong,[2] TW Ho,[1] PC Tong,[1] WM Lai,[1] ALT Ma[1]

[1]Paediatric Nephrology Centre, Hong Kong Children's Hospital; [2]Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

Background and Purpose: To evaluate the demographics and outcomes among children with End-stage kidney disease (ESKD) in Hong Kong.

Methods: We conducted a cohort study at the Paediatric Nephrology Centre, the designated site providing kidney replacement therapy (KRT) for children in Hong Kong. All children who initiated chronic KRT before 19 years, between 2001-2020, were included. Demographics, trend and survival outcomes of ESKD were examined.

Findings: 147 children (50% male) received KRT at a mean age of 11.4±5.7 years. The incidence of ESKD was 6.28 per million age-related population (pmarp). The leading cause of ESKD was congenital anomalies (33%). Ten children (7%) had pre-emptive kidney transplants, 104 (71%) and 33 (22%) patients received automated peritoneal dialysis and haemodialysis as initial KRT. The incidence of ESKD increased over time, and were 4.38, 5.07, 6.15 and 9.17 pmarp during 2001-2005, 2006-2010, 2011-2015 and 2016-2020, respectively (p=0.005). The mortality rate was 9.1 deaths per 1000-patient-years (95% CI 4.6-16.2). The survival probabilities at 1-, 5-, 10- and 15-year were 100%, 94.8% (95% CI 90.7-98.9%), 89.7% (95% CI 83.4%-95.9%), 87.1% (95% CI 79.3%-94.9%), respectively. Standardised mortality ratio was 54.5. >70% of deaths were due to infections. Young infants and those without kidney transplants were associated with worse survival (ps<0.01). Multivariate analysis demonstrated that patients receiving dialysis only had a significantly higher risk of death (HR_{adj} 12.9, 95% CI 2.7-63.2, p=0.002).

Conclusion: There is an increasing incidence of paediatric ESKD in Hong Kong. Mortality risk is comparable to other developed countries and is highest among dialysis population.

Recto-Urethral Fistula with a Normal Anus in a Boy with Vacterl Association: A Case Report and Literature Review

SLH Cheung, JWS Hung, KLY Chung, MWY Leung

Department of Surgery, Hong Kong Children's Hospital, Hong Kong SAR

Anorectal malformation (ARM) is a congenital malformation that has an incidence of 1 in 5000. It is classified into different types according to the Krickenbeck classification. Recto-urethral fistula with a normal anus is a rare variant among all ARM with very few cases reported in previous literature. We hereby report a case with H type recto-urethral fistula with a rare clinical presentation of recurrent epididymo-orchitis, with our subsequent workup, surgical management and literature review.

Impact of School Closure on Children'S Sleep Pattern During the COVID-19 Pandemic

GT Chua, WHS Wong, HK So, KM Yip, T Lam, WWY Tso, P Ip

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

Background: School closure is one of the main global health policies performed worldwide during the coronavirus disease 2019 (COVID-19) outbreak. Despite all of the advantages, there may be some risks for children who are quarantined. This study aimed to objectively measure and compares the sleep patterns of Hong Kong school students before and during the COVID-19 outbreak.

Methods: Baseline assessment was performed before the first wave of the COVID-19 outbreak in Hong Kong. The sleep pattern was recorded by a physical activity monitor (Actigraph wGT3X-BT, Pensacola, Florida, USA). The follow-up assessment was conducted in early 2020.

Findings: In total, 718 students were collected in the baseline. Subsequently 140 students joined the reassessment between March and April 2020. Analysis of sleep timing shows that 98.0% primary students, 78.0% secondary students and 79.9% primary school students and 58.8% secondary school students go to bed before midnight before and after the outbreak, respectively (p<0.001). Mean sleep duration (hours) was 6.81 (0.62) and 8.09 (0.07) at baseline and during the outbreak, respectively. The differences in the mean sleep features for total sleep time,

sleep fragmentation index and sleep fragmentation were 0.92 (1.64), 1.64 (6.95) and 2.49 (9.18), respectively. The overall sleep quality was poorer as evidenced by delays in bedtime and wake up time, increased duration in bed, longer sleep latency, increases in the movement and fragmentation indices.

Conclusion: This exceptional longitudinal study reported objective data on the change in sleep patterns before and during the COVID-19 outbreak with school closures.

A Chinese Boy with a Novel Compound Heterozygous Mutation of the DGKE Gene and Membranoproliferative Glomerulonephritis

SHY Lau,[1] EYH Chan,[2] LYP Yuen,[3] WF Ng,[3] ALT Ma[2]

[1]Department of Paediatrics, Prince of Wales Hospital; [2]Paediatric Nephrology Centre, Department of Paediatrics, Hong Kong Children's Hospital; [3]Department of Pathology, Hong Kong Children's Hospital, Hong Kong SAR

Background: Diacylglycerol kinase epsilon (DGKE) gene mutation is known to cause atypical haemolytic uremic syndrome. With its gene pleiotropy, a small number of patients were found to present with isolated membranoproliferative glomerulonephritis (MPGN).

Method: We report a ten-year-old Chinese boy with steroid-resistant nephrotic syndrome and a pathological diagnosis of immune-complex mediated MPGN. He first presented at the age of four with generalised oedema, hypoalbuminaemia, nephrotic-range proteinuria (urine pr/cr 13.9 mg/mg) and hyperlipidaemia. He responded poorly to full dose prednisolone and renal biopsy confirmed immune-complex mediated MPGN. He attained partial remission eventually with the use of Cyclosporin A. Four years later, he developed urinary relapse (urine pr/cr 1.57 mg/mg) after discontinuation of immunosuppressants.

Findings: Next-generation sequencing revealed compound heterozygous mutations in the DGKE gene. These included a c.1068_1071del p.(Asn356Lysfs*6) frameshift mutation and a c.1282_1284+18del deletion, both were classified as pathogenic (ACMG/AMP classification). Repeated renal biopsy showed immune-complex mediated MPGN and features of mesangiolysis, suggestive of previous glomerular thrombotic microangiopathy (TMA). This might represent a previous episode of subclinical TMA related to the DGKE mutations, in addition to an immune mediated process. Our patient showed good response to angiotensin converting enzyme inhibitor, prednisolone and mycophenolate mofetil and is now in complete remission.

Conclusion: This is the first Chinese patient of DGKE nephropathy presenting as nephrotic syndrome with MPGN picture. These two DGKE pathogenic mutations are also first to be reported among DGKE-patients with MPGN. Of interest, our patient responded promptly to immunosuppressants. This case highlights the importance of genetic testing in children with atypical course of nephrotic syndrome.

The Hidden Costs of Rare Diseases Beyond Healthcare Setting

CCY Chung, YNC Ng, ACY Lui, JLF Fung, MCY Chan, WHS Wong, SL Lee*, BHY Chung*

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

*Co-corresponding authors

Background: In Hong Kong (HK), one in 67 people is living with at least one rare disease (RD). While the impact is often perceived as the immediate healthcare burden, the broader socio-economic consequences of RDs are important for healthcare and related planning, yet challenging to estimate.

Purpose: To estimate the socio-economic cost of RDs in HK.

Methods: The Client Service Receipt Inventory for the RD population (CSRI-Ra) was used to collect direct and indirect cost-related data in the RD population in HK. RD patients and carers were recruited via multiple RD organisations in Hong Kong. Costs were estimated from the societal perspective using a bottom-up approach.

Findings: A total of 286 independent participants were recruited between April and August 2020, covering 106 unique RDs. Over 80% of RD patients (n=230) have utilised hospital healthcare services in the reported period; with an average of 36.6 inpatient days, 9.5 outpatient attendances, 3.6 accident and emergency visits, 10.2 day-care attendances, and 31.0 allied health visits per year among those who utilised the respective service. Of those who utilised residential medical services, there was an annual average of 29.7 visits per patient. Total hospital and community health service costs was found to be $25,703,312. A total of 176 patients (61.5%) took medications over the reported period, with an average of

5.5 drugs per patient (range: 1-27), equivalent to an annual total cost of $43,433,933. Over 70% (n=210) of RD patients required care from a paid or unpaid carer. Annual costs of paid care and informal care support were found to be $4,281,545 and $36,834,569, respectively. Due to the patient's RD condition, labour productivity loss in the form of annual leave and reduced working hours was equivalent to an annual cost of $28,577,883. The total annual cost for the 286 RD patients was estimated to be a minimum of HKD$138,831,242, with a minimum average cost of HKD$485,424 per patient per year.

Conclusion: This is the first study to estimate the socio-economic costs of RDs in the healthcare and community settings in HK. Hidden costs in the form of informal care support and productivity losses are significant in this population, reflecting the importance of rare diseases in health policies.

Acknowledgement: *We would like to thank the Society for the Relief of Disabled Children for their funding support, and all patients and carers who participated in the study.*

Theory of Mind (ToM) Profiles in Children with Autism Spectrum Disorder (ASD) and Developmental Language Disorder (DLD)

CN **Hung**, CCC **Lam**, EKF **Woo**, SKY **Liu**, CS **Sheh**, L **Lam**, SP **Cheung**

Child Assessment Service, Department of Health, Hong Kong SAR

Background/ Introduction: Theory of Mind (ToM) is a fundamental social cognitive skill necessary for understanding of intentions, emotions, desires, taking perspective and making predictions about other's behaviour.

ToM is a composite function requiring visual/auditory input, language, central coherence and executive function and may be affected in different neurodevelopmental disorders such as autism spectrum disorder (ASD), developmental language disorder (DLD), attention deficit hyperactivity disorder (ADHD), hearing impairment and visual impairment.

The Hong Kong Scales for Assessment of Theory of Mind (HKAToM) was published by the Child Assessment Service (CAS) in 2020 for evaluating ToM of children aged 5 years to 12 years 1 month.

Purpose: To review HKAToM performance profiles in 85 children with suspected language impairment or ASD in CAS from 2019 to 2020.

Method: (1) Language abilities and ToM performance was compared in 35 children with suspected language impairment aged 5-12 years. (2) ToM abilities were analysed in 50 children with ASD and normal intelligence aged 6-12 years.

Findings: (1) Among children with suspected language impairment, there was a significant correlation between total score of HKAToM and specific subtests of Hong Kong Cantonese Oral Language Assessment Scale (HKCOLAS). (2) Among children with ASD, performance on basic ToM tasks was comparable to norm but they presented significant difficulty in more advanced ToM tasks.

Conclusions: Language impairment is correlated with poor ToM. In children with ASD, compensatory reasoning supported basic but not advanced ToM tasks. These findings provide useful information for understanding the social interaction difficulties of these children, and for guiding their training.

Volume-Targeted Versus Amplitude-Targeted High Frequency Oscillatory Ventilation: A Retrospective Case Control Study

HCK **Yau**, CSB **Cheung**, SK **Yip**, HSHS **Lam**

Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR

Background: High-frequency oscillatory ventilation (HFOV) is widely used in neonatology. Fluctuations in pCO2 increase risk of intraventricular haemorrhage and hypocarbia is associated with cerebral ischaemia in preterm infants. Carbon dioxide (CO2) elimination during HFOV correlates with the diffusion coefficient of CO2 which is very sensitive to changes in tidal volume. Volume-guaranteed HFOV (HFOV-VG) is an advanced ventilation mode that allows clinicians to target the tidal volume delivered rather than the amplitude, potentially reducing pCO2 fluctuations.

Purpose: To retrospectively evaluate the difference in pCO2 variability between preterm neonates receiving HFOV-VG and conventional HFOV.

Methods: Preterm infants who received HFOV-VG in the Prince of Wales Hospital were identified. Each infant who received HFOV-VG was gestational age-, birth weight- and sex-matched with two infants who received conventional HFOV. The mean and standard deviation (s.d.) of pCO2 levels, and percentages of hypercarbic and hypocarbic readings were compared using Mann-Whitney U test and unpaired t-test as applicable.

Findings: Results showed significant difference in the pCO2 s.d. between the HFOV-VG and conventional HFOV groups (p-value=0.0451; mean difference=0.496). Incidence of hypocarbia (pCO2 <4 kPa) was significantly lower in the HFOV-VG group (p-value=0.002).

Conclusions: Preterm neonates receiving HFOV-VG had less fluctuations in pCO2 and lower incidence of hypocarbia than those receiving conventional HFOV. It suggests that HFOV-VG might be preferred in preterm neonates who are susceptible to cerebral vascular injuries. To confirm our findings, larger-scale randomised controlled trials are needed.

Paediatric Adrenocortical Tumours in Hong Kong - 25 Years of Experience

CLS Yan, SLK Lee, JYL Tung, DTL Ku, ECH Fu, JPW Yau, MMK Shing, CW Luk, GCF Chan, APY Liu

Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Background: Adrenocortical tumours (ACTs) are rare neoplasms in children encompassing benign adenoma (ACA) and malignant carcinoma (ACC). Endocrinologic dysregulation is frequent and underlying cancer predispositions have to be considered.

Purpose: To review the characteristics and treatment outcome of paediatric ACTs in Hong Kong leveraging a population-wide childhood cancer database.

Methods: Retrospective review of the Hong Kong Paediatric Haematology/Oncology Study Group database for patients diagnosed <18 years-old with ACTs from May 1996 to April 2021.

Findings: Thirteen patients with ACTs were identified (ACC=11, ACA=2; M:F=6:7). Median age of diagnosis was 8.2 years (range: 6 months-15 years). Most patients (n=8) presented with virilisation or precocious puberty, whereas hypertension (n=3), abdominal pain (n=3), hypokalemia (n=1) were less common features. Biochemically, excessive androgen (n=10) was most frequent, followed by cortisol (n=3) and aldosterone (n=1) excess. COG staging was I in 6 patients, II in 3, III in 1 and IV in 3. Twelve patients received surgical resection of primary tumour which was preceded by biopsy in 4. Chemotherapy was given in 5 patients (Stage I=1, III=1, IV=3) using cisplatin/etoposide/doxorubicin with mitotane. All 5 patients progressed among which 4 died within 2 years despite further second-line treatment. In contrast, all 8 patients (Stage I/II disease) who had resection alone remained disease-free (median follow-up 9.8 years). Germline *TP53* mutations were detected in 4/7 patients (pathogenic=3, variant of unknown significance=1) tested; one patient had Beckwith-Wiedemann syndrome.

Conclusions: The management of ACT requires multi-disciplinary input and advanced-stage disease remains a challenge direly requiring novel strategies.

Hearing Impairment in Children with Waardenburg Syndrome

WM Chan, CC Lam, PW Yip, CP Chow

Child Assessment Service (CAS), Department of Health, Hong Kong SAR

Background and aims: Waardenburg syndrome (WS) is a rare condition characterised by hearing loss and pigmentation deficiencies. Other anomalies vary depending on the type. This study aims to review the hearing impairment (HI) in children with WS and their developmental profiles.

Methods: CAS data was collected on children born from 2013 to 2020 with WS and significant HI. Reasons for referral, hearing thresholds, use of hearing aids or cochlear implantations (CI) and developmental findings were analysed.

Results: Six cases were identified. The hearing ranged from moderate to profound sensorineural hearing loss. The developmental profile ranged from mild language delay to mild intellectual disability.

Four cases of HI were detected at universal newborn hearing screening (UNHS) and confirmed with significant HI.

The remaining two children presented with bilateral blue iridis without dystopia canthorum. There was no confirmed HI at referral to CAS. One child presented at 2 years 2 months for developmental delay and suspected autism spectrum disorder. Profound SNHL in one ear and severe SNHL in the other were diagnosed. The second child presented at 1 year 3 months for language delay, hypotonia and gross motor delay. Bilateral severe to profound SNHL was found. CIs were done for both children, which improved hearing thresholds to conversational levels.

Conclusions: We should be alert of WS and consider hearing assessment for children with bilateral blue iridis without dystopia canthorum.

Study on the Mucosal and Serological Immune Response to the Novel Coronavirus(SARS-CoV-2) Vaccines

RWY Chan,[1,2,3,4] SJ Liu,[1,2,3,4] JY Cheung,[1,2,3,4] JGS Tsun,[1,2,3,4] KC Chan,[1,2] KYY Chan,[1,2] GPG Fung,[1] AM Li,[1,2,4] HS Lam[1,2]

[1]Department of Paediatrics, Faculty of Medicine; [2]Laboratory for Paediatric Respiratory Research, Li Ka Shing Institute of Health Sciences, Faculty of Medicine; [3]CUHK-UMCU Joint Research Laboratory of Respiratory Virus & Immunobiology, Department of Paediatrics, Faculty of Medicine; [4]Hong Kong Hub of Paediatric Excellence, The Chinese University of Hong Kong, Hong Kong SAR

Vaccines that elicit mucosal immune responses against SARS-CoV-2 could potentially be of exceptional importance in providing first line defence at the site of viral entry. In order to understand the mucosal immune response profiles of SARS-CoV-2 vaccines, we examined both the mucosal and systemic responses of subjects vaccinated by two different vaccination platforms: mRNA (Comirnaty) and inactivated virus (CoronaVac). Nasal epithelial lining fluid (NELF) and peripheral blood samples were collected in subjects who had received two doses of CoronaVac or Comirnaty. We quantified IgA and IgG specific to SARS-CoV-2 S1 protein, neutralisation antibody by ELISA in NELF and plasma samples. Only Comirnaty induced nasal S1-specific IgA and IgG responses, which were evident as early as on 14±2 days after the first dose. The NELF samples of 72% of subjects became IgA+IgG+, while in 62.5% of subjects the samples were neutralising by 7±2 days after the second dose. In 45% of the subjects their NELF remained neutralising 50 days after the booster. In plasma, 91% and 100% Comirnaty subjects possessed S1-specific IgA+IgG+ on 14±2 days after the first dose and 7±2 days after booster, respectively. The plasma collected on 7±2 days after booster was 100% neutralising. The induction of S1-specific antibody by CoronaVac was IgG dominant, and 70% of the subjects possessed specific IgG by 7±2 days after booster and were all neutralising. This study reveals that Comirnaty is able to induce S1-specific IgA and IgG response with neutralising activity in the nasal mucosa in addition to a consistent systemic response.

Predictors for Asthma Persistence in a Longitudinal Cohort of Chinese Asthmatic Children

JCH Leung,[1,2] MF Tang,[1,3] ASY Leung,[1] TF Leung[1,3]

[1]Department of Paediatrics, Prince of Wales Hospital; [2]Faculty of Science and [3]Hong Kong Hub of Paediatric Excellence, The Chinese University of Hong Kong, Hong Kong SAR

Background: It is a common experience that some children with asthma outgrow their airway disease with time whereas a small proportion of them have persistent asthma. There is limited evidence on factors associated with persistence of childhood asthma in Asian populations.

Objective: To identify predictors for asthma persistence in Hong Kong Chinese children.

Methods: A cohort of 181 asthmatic children aged 5-15 years from paediatric allergy clinic of our university-affiliated teaching hospital were prospectively followed by the same paediatrician for at least 5 years and into their early adulthood. Logistic regression was used to identify independent factors associated with asthma persistence.

Results: The mean (SD) age of patients at recruitment was 10.0 (2.7) years, and they were followed for 12.5 (2.8) years. Sixty-one percent had two or more episodes of asthma exacerbation, 64% received inhaled corticosteroid (ICS) treatment ever, and 34% had outgrown asthma. Boys were more likely to outgrow asthma (odds ratio [OR] 2.50 and 95% confidence interval [CI] 1.19-5.23) and the younger the patients were at the initial visit, the more likely they outgrew asthma (OR 1.15 and CI 1.01-1.32). On the other hand, children who received ICS treatment ever and those with two or more episodes of asthma exacerbations were more likely to have persistent asthma (OR 3.70 and CI 1.78-7.68; OR 2.59 and CI 1.28-5.26).

Conclusions: Boys and younger age at baseline were more likely to outgrow asthma while ICS treatment ever and history of frequent asthma exacerbations are predictors for persistent asthma into early adulthood.

Innate Immune Response of Human Influenza a Virus and Rhinovirus A16 in Patients' Nasopharyngeal Aspirates and Human Airway Models

BKS Wong,[1,2,3,4] JGS Tsun,[1,2,3,4] LLY Chan,[1,2,3,5] KP Tao,[1,2,3,4] X Long,[1,2] SMW Chow,[6] CSH Ng,[7] RWH Lau,[7] TF Leung,[1,2,3,4] AM Li,[1,2,3] RWY Chan[1,2,3,4*]

[1]Department of Paediatrics, Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR; [2]Hong Kong Hub of Paediatric Excellence, The Chinese University of Hong Kong, Hong Kong SAR; [3]CUHK-UMCU Joint Research Laboratory of Respiratory Virus & Immunobiology, Department of Paediatrics, Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR; [4]Laboratory for Paediatric Respiratory Research, Li Ka Shing Institute of Health Sciences, Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR; [5]Lee Kong Chian School of Medicine, Nanyang Technological University, Singapore; [6]Department of Otorhinolaryngology, Head and Neck Surgery, The Chinese University of Hong Kong, Hong Kong SAR; [7]Department of Surgery, The Chinese University of Hong Kong, Hong Kong SAR

*Corresponding author

Background/Introduction: Rhinovirus (RV) and influenza A virus (IAV) are prevalent respiratory viruses found in children. RV infection is considered to induce a milder response than IAV in general. Whether the parameters measured in the mucosal fluid of patients can reflect by the in vitro cell models remain elusive.

Purpose: To compare the mucosal inflammatory profiles of hospitalised RV- and IAV-positive children and verify if the air-liquid interface (ALI) culture of the human nasopharyngeal (NP) and bronchial epithelia-derived epithelial cell culture and the corresponding airway organoids (AOs) are good models to reflect the local immune response.

Methods: Nasopharyngeal aspirates (NPA) from hospitalised paediatric patients with respiratory symptoms are collected during admission. The presence of respiratory virus was detected by multi-plex PCR and RV was genotyped by VP4-VP2 sequencing. The cell pellet of the NPA was extracted for measuring host gene expressions. ALI and AO cultures were cultured from human NP and bronchial tissue, and infected with RV-A16 and IVA H1N1. Viral replication and gene induction were assessed.

Findings: A total of thirteen RV-A and fifteen IAV positive paediatric subjects were identified. RV-A patients were significantly younger than IAV patients. Influenza virus induced a stronger chemokine response in patients' NPA, including CCL8, CXCL11 and CCL5 than RV-A, as detected in their NPA. IAV may induce stronger IFN response in ALI and AO culture.

Conclusion: We identified distinct innate immune responses of RV-A and IVA in patients' NPA and human airway models. AOs serve as a novel model for IVA pathogenesis.

Teamwork on Early Mobilisation for a Critically Ill Patient on Continuous Renal Replacement Therapy (CRRT) in the Paediatric Intensive Care Unit (PICU) - A Case Study

CH Pak,[1] WF Hui,[2] YB Ho,[1] NC Chan,[1] KL Hon[2]

[1]Allied Health Department (Physiotherapy); [2]Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Recent studies showed early mobilisation during CRRT in PICU is safe to minimise the risk of deep vein thrombosis (DVT) and alleviate patients' post-PICU symptoms. This case study demonstrates the teamwork in PICU to achieve early mobilisation for a critically ill patient on CRRT.

Methodology: A 7-year-old boy with hepatoblastoma and lung metastasis was admitted to the PICU. His condition deteriorated with ventilator support, CRRT and latter intermittent haemodiafiltration (IHF). Medical and physiotherapy records were reviewed. Children's Chelsea Critical Care Physical Assessment Tool (cCPAx) and maximum inspiratory and expiratory pressures (MIP & MEP) were measured. Any adverse effect were documented.

Results: Chest physiotherapy were practiced since admission. Inspiratory muscle training via endotracheal tube (ETT) initiated to promote the respiratory function (intubated MIP and MEP: 20 cmH_2O). Anti-embolism stockings accompanied with mobilisation were administrated.

During the CRRT, an active video gaming incorporated into rehabilitation promoted exercise compliance. Strategies for early sit out of bed (SOOB) were discussed in weekly PICU round i.e., IHF catheter was repositioned into right internal jugular vein. SOOB for IHF successfully achieved since day10.

Since extubation on day18, assisted walking and tricycling were practiced with improvement at MIP and MEP (31 and 33 cmH_2O on day20) and cCPAx total score (from 2/50 (admission) to 35/50 (day 45)). No adverse event was reported.

Conclusion: Teamwork between the PICU team and physiotherapists provides a safe example for early mobilisation for PICU care with CRRT. This serves a platform for further development of early mobilisation protocol in PICU.

A Challenging Case for Genetic Counselling: Blended Phenotype of Familial Hypocalciuric Hypercalcaemia, Haemophilia A and Turner Syndrome with Interaction Between Isochromosome X and Duplication in *F8*

PHL **Doong**, JFT **Chau**, CCY **Mak**, MHY **Tsang**, JLF **Fong**, CCY **Chung**, MMC **Chui**, M **Lee**, AKY **Kwong**, BHY **Chung**

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

Introduction: Blended phenotype, a complex phenotype affected by more than one defective gene, is not as rarely seen as it used to be because of the advancement of whole exome sequencing. Retrospective analysis of WES data of 7374 patients shows 4.9% of patients were diagnosed with two or more monogenic diseases (Posey et al., 2016).

Case Report: This case report describes a patient with a blended phenotype of familial hypocalciuric hypercalcaemia, type I (FHH1), haemophilia A and Turner syndrome. At age of 1 year old, she was noticed to have spontaneous bruising with abnormal clotting. Blood test revealed she has more severe haemophilia A, with factor VIII <1% clotting activity, than her father. Constitutional karyotyping was performed to determine the genetic cause of the severe expressivity of haemophilia A and identified an Xq11.1-28 duplication affecting the *F8* gene in addition to the *F8* genetic defect inherited from her father in another X chromosome. Besides, mosaic Turner syndrome was substantiated.

Single gene testing of *CASR* was arranged because of positive family history and found a paternally inherited heterozygous mutation (NM_000388.4:c.196C>T, p.Arg66Cys) in *CASR* associated with FHH1, an autosomal dominant disease requiring no treatment but monitoring.

Conclusion: This is a challenging case for genetic counselling because it is uneasy to educate the patient about the modes of inheritance of the three distinct genetic diseases. Since the two defective genes segregate independently, a dihybrid cross model has to be used to explain the integrated recurrence risk. There were also difficulties in the diagnostic journey and provision of preimplantation genetic testing to the family.

Associations Between Childhood Maltreatment and Psychiatric Disorders: Findings from a Population-Based Cohort Study

RS **Wong**,[1] KTS **Tung**,[1] FKW **Ho**,[2] TMC **Lee**,[3,4] KL **Chan**,[5] J **Bacon-Shone**,[6] D **Coghill**,[7,8] KKC **Man**,[9,10] PC **Sham**,[11,12] WHS **Wong**,[1] WWY **Tso**,[1] GT **Chua**,[1] ICK **Wong**,[9,10] P **Ip**[1]

[1]Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR; [2]Institute of Health and Wellbeing, University of Glasgow, United Kingdom; [3]State Key Laboratory of Brain and Cognitive Sciences, The University of Hong Kong, Hong Kong SAR; [4]Laboratory of Neuropsychology, The University of Hong Kong, Hong Kong SAR; [5]Department of Applied Social Sciences, The Hong Kong Polytechnic University, Hong Kong SAR; [6]Social Sciences Research Centre, The University of Hong Kong, Hong Kong SAR; [7]Department of Paediatrics and Psychiatry, Faculty of Medicine, Dentistry and Health Sciences, University of Melbourne, Melbourne, Australia; [8]Murdoch Children's Research Institute, Melbourne, Australia; [9]Centre for Safe Medication Practice and Research, Department of Pharmacology and Pharmacy, The University of Hong Kong, Hong Kong SAR; [10]Research Department of Practice and Policy, UCL School of Pharmacy, London, United Kingdom; [11]Department of Psychiatry, The University of Hong Kong, Hong Kong SAR; [12]Centre for PanorOmic Sciences, The University of Hong Kong, Hong Kong SAR

Background: High-quality evidence regarding links between childhood maltreatment and psychiatric diagnoses is lacking particularly for Axis II disorders.

Purpose: This study used electronic health record data to explore the association between childhood maltreatment experiences and Axis I and Axis II mental disorders.

Methods: In this study, the exposed group (n=7,473) comprised patients aged 0 to 19 years with a first-time record of maltreatment episode between January 1, 2001 and December 31, 2010, whereas the unexposed group (n=26,834) comprised individuals with the same gender, age, and admission hospital and time who did not have any record of maltreatment episodes in the Hong Kong Clinical Data Analysis and Reporting System (CDARS). Data on their psychiatric diagnoses recorded between the date of admission and January 31, 2019 were also retrieved. A Cox proportional hazard regression model was fitted to estimate the hazard ratio (HR, plus 95% CIs) between childhood maltreatment exposure and psychiatric diagnoses, adjusting for age at index visit, sex, and government welfare recipient status.

Findings: Childhood maltreatment exposure was significantly associated with subsequent diagnosis of conduct disorder/oppositional defiant disorder (adjusted HR, 10.99 [95% CI 6.36, 19.01]), attention deficit

hyperactivity disorder (ADHD) (7.28 [5.49, 9.65]), and personality disorders (5.36 [3.78, 7.59]). The risk of subsequent psychiatric disorder in maltreated children did not vary by exposure to sexual abuse, age at exposure, and gender.

Conclusions: Maltreated children are vulnerable to psychiatric disorders. Findings highlight the need for early provision of integrated family support services to address the long-term psychosocial needs of maltreated children.

Association Between Chronic Inflammation and Telomere Length Among Overweight and Normal-Weighted Adolescents

KTS **Tung**, RS **Wong**, HW **Tsang**, CHY **Chow**, P **Ip**

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR

Background: Rising prevalence of obesity in children and adolescents over the past decades has become a worldwide health concern. Recent research found that overweight and obese children and adolescents are more likely to have shorter telomere length (TL), which is recognised as one of the biomarkers of ageing. However, the biological pathways underlying these associations remain to be unknown.

Purpose: The purpose of this study is to investigate the association between serum cytokine levels and telomere length among overweight and normal-weighted adolescents.

Methods: A total of 278 adolescents aged 12 to 15 years were included in the analyses of this study. Buccal swabs and peripheral venous blood samples were collected from the adolescents for measuring telomere length and serum cytokines levels respectively. Anthropometric measurements on parameters including weight and height in barefoot and light clothing were also obtained.

Findings: Among the 278 participants, 180 were boys (64.7%) and 98 were girls (35.3%) with an average age of 13.35 years. About one-third of the adolescents (31.3%) were overweight with the average BMI of 19.97. Adjusted regression models showed that the levels of interleukin (IL)-1β, tumour necrosis factor (TNF)-α, IL-6, and IL-8 were significantly associated with TL. Only IL-18 and IL-1β were significantly associated with TL in normal-weighted adolescents.

Conclusions: This study adds to the current evidence that TL among overweight adolescents are more vulnerable to the elevated levels of peripheral monokines. This indicates the potential involvement of macrophages in the process of cellular aging, especially among obese and overweight population.

Bedside Surgical Release of Tongue Tie in Babies with Breastfeeding Difficulties: A Report on the Local Experience

KKY **Pang**,[1] JWC **Mou**,[1] CK **Chan**,[2] KW **Lai**,[2] CCO **Lam**,[3] SK **Wong**[3]

[1]Division of Paediatric Surgery and Paediatric Urology, Department of Surgery, Prince of Wales Hospital; [2]Department of Obstetrics and Gynaecology, Prince of Wales Hospital; [3]Department of Obstetrics and Gynaecology, Queen Elizabeth Hospital, Hong Kong SAR

Background: Tongue tie is a common anatomical variation found in 1.7-10% of the population. While it seldom carries functional impairment into older age, there were studies showing babies with tongue tie experience more difficulties with direct breastfeeding. A few randomised controlled trials suggested the benefit of surgical release of tongue tie in this context. These had led to the recommendation from National Health Service and the Canadian Pediatric Society on the surgical procedure. In Hong Kong, despite increasing awareness on the benefits of breastfeeding, a comprehensive service is yet to be established for babies encountering difficulty in breastfeeding associated with tongue tie.

Purpose: We report our experience in the surgical release of tongue tie in conjunction with assessment and review by lactation consultants.

Method: A retrospective review was carried out in babies with surgical release of tongue tie who presented with difficulty with breastfeeding difficulties. Preoperative complaints were documented. All of them had reassessment on breastfeeding by a lactation consultant immediately after the procedure.

Findings: In the study period of 26 months, a total of 46 babies had bedside release of tongue tie performed in Prince of Wales Hospital. Forty-one (89%) of them had pre-operative and post-operative assessment by lactation consultants. Presenting complaints included ineffective sucking/slow feeding (30%), poor latching (61%) and sore nipples (37%). No complication has been raised from the procedures.

All babies were immediately assessed for breastfeeding after the procedure. All of them had the symptoms

improved. Only 30 babies were followed up after the procedure day, at a median follow up of 8 days. Twenty-six (87%) were continuing direct breastfeeding. Thirteen mothers had a delayed interview on the scale of improvement (rated 0-10) from the procedure, mean score was 8.7.

Conclusions: In the local setting, surgical release of tongue tie at bedside brought immediate improvement in the symptoms from tongue tie in breastfeeding babies. A standardised follow up practice is needed to study the longer term effect of the procedure.

Effects of an Evidence-Based Educational Programme for Improving Nurses' Knowledge and Self-Efficacy About the Ponseti Method of Clubfoot Treatment: Service Innovative Project

KM Chui, WY Lee

Department of Paediatric and Adolescent Medicine, United Christian Hospital, Hong Kong SAR

Background: Clubfoot, also known as congenital talipes equinovarus, is one of the most common deformities of the lower limbs in newborns. It is treatable and can be corrected by Ponseti method with 90% of successful rates. However, relapse is the most common problem affecting long term successful outcome. Poor adherence with Ponseti method is regarded as the major cause of relapse. A lack of knowledge and skills about Ponseti method and misunderstanding about the importance of brace compliance among healthcare providers and family caregivers of clubfoot children are the common cause. Hence, researchers suggested that relevant training or education should be provided to health care providers so that they can acquire the knowledge and skills of clubfoot management. Unfortunately, relevant training for health care providers were mostly provided in low- and middle-income countries and there is insufficient education or training about clubfoot management in Hong Kong. Also, the admission rate of clubfoot is increasing in these few years so there is a need to implement evidence-based educational programme for nurses to improve the knowledge and self-efficacy about the Ponseti method of clubfoot treatment.

Aims & objectives: The aim of this service innovative project is to develop and pilot testing an evidence-based educational programme for nurses who take care of clubfoot children undergoing Ponseti method of treatment. Specific objectives are: (1) to evaluate the effectiveness of the educational programme on nurses' knowledge and self-efficacy related to clubfoot and Ponseti method of clubfoot treatment; and (2) to evaluate the level of satisfaction and feasibility of the educational programme among nurses.

Potential setting: The educational programme will be conducted in an indoor seminar room in the hospital.

Potential participants: Nurses, regardless of age and clinical experiences, who are currently working in the paediatric wards and need to take care of children with clubfoot in an acute public hospital will be recruited.

Project plans: The educational programme involves two face-to-face education sessions taught by an orthopaedic surgeon and an orthopaedic nurse consultant, who are experienced in Ponseti method. There will be two face-to-face education sessions in two consecutive weeks. Each face-to-face education session will last for around 3 hours. Two face-to-face education sessions include theoretical and practical sessions. Through theoretical sessions, knowledge about clubfoot and Ponseti method of clubfoot treatment will be delivered to nurses through lectures. Each nurse will receive a booklet which comprised of lecture notes with powerpoint slides. Through practical sessions, skills of cast and brace application will be demonstrated to nurses and they need to return demonstration on cast and brace application. Question & answer session and group discussion are also provided after each education session.

Project outcomes: Demographic data of nurses will be collected before the start of educational programme. Also, primary outcome is to assess nurses' knowledge of clubfoot and Ponseti method. Secondary outcome is to assess nurses' self-efficacy regarding to take care of clubfoot children undergoing Ponseti method. Outcomes will be measured before and after the implementation of intervention.

Discussion: This educational programme is feasible in applying to clinical settings due to its high degree of practicality, implementation and demand. The cost of education materials and settings are relatively low. Also, it is facilitated and supported by Department of Paediatrics and Adolescent Medicine. Flexible arrangement of duty can be done to encourage nurses to participate in the training.

Conclusion: Through the service innovative project, nurses are expected to increase the knowledge and self-efficacy to take care of the clubfoot children during different phases of Ponseti method of treatment. Thus, they can provide appropriate care and education to patients and family caregivers to increase their satisfaction of treatment,

enhancing patient outcomes and finally improving the quality of health care service about clubfoot in our working paediatric unit.

ESS is an Independent Predictor of Systolic Blood Pressure in Sleep-Deprived Schoolchildren

E Cheng, M Ng, KCC Chan, CT Au, AM Li

Department of Paediatrics, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong SAR

Introduction: In Hong Kong, sleep deprivation is common amongst schoolchildren. While it is associated with numerous psychosocial and health consequences, for example worse daytime functioning, emotional disturbances and obesity, recent studies also evaluated its impacts on systolic blood pressure (SBP) in the paediatric population. Various cohorts have reported higher SBP in sleep-deprived schoolchildren when compared to those who have sufficient sleep. However, the predictors of a higher SBP in sleep-deprived subjects remain unclear.

Purpose: A cross-sectional study was conducted to assess the potential predictors of SBP in Hong Kong schoolchildren.

Methods: 53 sleep-deprived subjects aged between 12-16 were included after excluding those with obesity, sleep disorders and any known conditions or medications that may affect sleep. Their body weight, height, BMI, pulse rate, blood pressure and sleep duration were recorded. The Paediatric Daytime Sleepiness Scale (PDSS) and Epworth Sleepiness Scale (ESS) were completed at baseline. Sleep deprivation was defined as sleep duration <8 hours at baseline. The association between baseline characteristics and SBP was evaluated by simple linear regression. A backward multiple regression model was used to assess the effects of PDSS and ESS on SBP.

Findings: 53 sleep-deprived subjects with a mean sleep duration of 6.63 hours per day were included in our study. ESS [$F(1,51)=6.351$, $p=0.015$, $R^2=0.111$, $R^2_{adjusted}=0.093$], age [$F(1,51)$, $p=0.004$, $R^2=0.150$, $R^2_{adjusted}=0.133$] and BMI [$F(1,51)=4.607$, $p=0.037$, $R^2=0.083$, $R^2_{adjusted}=0.065$] are associated with SBP using simple linear regression. However, sleep duration in sleep-deprived schoolchildren is not associated with SBP [$F(1,51)=2.272$, $p=0.138$, $R^2=0.043$, $R^2_{adjusted}=0.024$]. Three independent variables, including ESS, age and BMI, were included in the multiple regression analysis [$F(3,49)$, $p<0.001$, $R^2=0.290$, $R^2_{adjusted}=0.247$]. The association between ESS and SBP remains statistically significant [$\beta=-0.336$, $p=0.010$]. Another multiple regression analysis was conducted, which included BMI, age and PDSS. PDSS is not associated with SBP in this multiple regression model [$\beta=-0.212$, $p=0.103$].

Conclusions: ESS is an independent predictor of higher SBP in sleep-deprived schoolchildren.

Photogrammetry as a Screening Tool for Childhood Obstructive Sleep Apnoea - A Pilot Study

HM Yuen, KCC Chan, AM Li, CT Au

Department of Paediatrics, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong SAR

Background: Cranio-facial profile is an important component in the aetiology of childhood obstructive sleep apnoea (OSA). Different craniofacial features can be captured by photogrammetry, which is making measurements by photographs. Whether photogrammetry findings together with clinical parameters would yield a clinically acceptable screening tool for OSA has not been explored.

Purpose: To develop a prediction model for childhood OSA using both clinical parameters and photogrammetric craniofacial features.

Method: Prepubertal children suspected of OSA were recruited. All subjects underwent craniofacial photogrammetry and overnight polysomnography. Data was split into 70% training data and 30% test data. The prediction models were built from the training data using logistic regression and evaluated on the test data using receiver operating characteristic curve analysis.

Results: This study included 90 children (mean age: 8.2±1.6 years, 66 males). Non-OSA, mild OSA and Moderate-to-severe (MS) OSA groups consisted of 32, 31, and 27 subjects, respectively. Four prediction models were built. Model 0 was built with only clinical measurements which included age, sex, BMI z-score and the presence of large tonsils as predictors (AUC=0.683). Models 1 and 2 used clinical measurements and one photogrammetric feature, which was the maxillary-mandibular relation angle (sn-n-sl) for model 1 (AUC=0.778), and the anterior mandibular height to whole face length ratio (sto-gn/n-gn) for model 2 (AUC=0.806). Model 3 used clinical measurements and the two photogrammetric features, giving the highest accuracy (AUC=0.861).

Conclusion: Craniofacial features obtained from photogrammetry could improve the prediction accuracy for childhood OSA.

Genetic Diagnosis of Mitochondrial Diseases by Detection of Aberrant Expression and Splicing Events in RNA Sequencing Data

AKY KWONG,[1*] JFT CHAU,[1*] VA YEPEZ,[2] M CHUI,[1] MHY TSANG,[1] M LEE,[1] CW FUNG,[1] BHY CHUNG[1]

[1]Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR; [2]Department of Informatics, Technical University of Munich, Munich, Germany

*With equal contributions

Background and objective: Mitochondrial diseases (MDs) are the commonest group of inborn errors of metabolism. Genetic diagnosis is crucial for disease management but challenging owing to phenotypic and genetic heterogeneity. MtDNA sequencing and whole exome sequencing (WES) are useful diagnostic tools, but a proportion of the patients remains undiagnosed. In this study, we aim to investigate the power of RNA sequencing (RNAseq) to overcome the limitation of WES for genetic diagnosis.

Methods: We studied a cohort of 25 undiagnosed patients with suspected MDs after WES. RNAseq was undergone for the fibroblasts of these 25 patients together with 6 genetic confirmed positive controls, 2 undiagnosed patients with other diseases and 8 unaffected controls. We implemented a recently established workflow, Detection of RNA outliers pipeline (DROP), to detect aberrant expression and splicing events.

Results: A total of 95 significant expression outliers and 1847 splicing outliers have been identified in the patients. Two significant aberrantly expressed genes, *MFSD1* and *GFM1*, were found in one of the undiagnosed MD patients. These two genes are located with high proximity on chromosome 3 and a possible deletion covering *MFSD1* is suggested. Further studies are needed to investigate whether the deletion will affect cis-acting elements controlling the expression of *GFM1* associated with oxidative phosphorylation deficiency. An aberrant splicing event was identified in another patient resulting in partial intron 2 retention in *POLRMT* encoding mitochondrial RNA polymerase. Functional study is essential to evaluate the effect of the intron retention on protein function.

Conclusion: Here we have investigated a transcriptome-directed approach for molecular diagnosis of MDs. Further investigation by alternative prioritisation of outliers and functional confirmation will be proceeded to identify candidate genetic defects to explain the patients' phenotypes.

Acknowledgements: We would like to thank The Society for the Relief of Disabled Children and HMRF Commissioned Paediatric Research Programme (PR-HKU-4) for the support.

Neurofibromatosis, Old and New: A 16 Years Clinical and Molecular Characterisation in Hong Kong

S HO,[#] HM LUK,[#] SYL NG, KPT YU, SSW CHENG, MTS MOK, EWL HAU, IFM LO[*]

Clinical Genetic Service, Department of Health, Hong Kong SAR

[#]Co-first authors; [*]Corresponding author

Background: Neurofibromatosis type 1 (NF1) is the commonest multi-systemic neurocutaneous tumour-predisposition disorder. It has an age-related complete penetrance but a highly variable inter- and intra-familial expressivity.

Purpose: To explore the incidences of features in the NIH clinical diagnostic criteria and the molecular characterisation in our cohort, report on novel variants and discover new genotype-phenotype correlations.

Methods: The clinical features and molecular characteristics of 832 clinically or molecularly confirmed NF1 patients from 697 unrelated families recruited from a single centre in Hong Kong diagnosed during the 16 years period from January 2005 to January 2021 were summarised and analysed.

Findings: While the majority of the reported figures in terms of clinical features and molecular findings appeared to be concordant with what had been described in the literature, we would like to highlight the newly found association of a heightened risk of congenital heart anomalies and pulmonary stenosis in individuals harbouring in-frame variants in the entire RAS-GAP domain, accompanied with a "Noonan-syndrome-like phenotype" with a higher incidence of short stature, relative macrocephaly, pectus abnormalities and lower incidence of cutaneous neurofibroma. On the other hand, the incidences of hypertension, stroke, epilepsy, vascular abnormalities and malignancies appeared much lower in our cohort than reported figures from previous literature, whether these findings reflect genuine ethnicity-related differences, or environmental factors and modifications present in our locality, requires further studies.

Conclusions: In this study, we have revisited the incidences of NIH clinical diagnostic criteria features and molecular characterisation, reported on novel variants and discovered new genotype-phenotype correlations.

Fathers' Involvement in Pregnancy and After Childbirth – A Qualitative Study

WI WONG

Union Hospital School of Nursing, Hong Kong SAR

Background: There was a significantly global trend in discussing fathers' experience and involvement in pregnancy and childbirth since 1960s. The number of fathers who were participated in engagement during pregnancy were also increasing. Some of the famous articles identified the importance of the presence of fathers' involvement such as attending antenatal care or cutting umbilical cord in delivery ward. In Hong Kong and Chinese culture, most of the family units are belonging to nuclear family. The tradition mother roles are full of domestic duties and mainly provide care to children. Mothers are playing a fundamental role in the children's life and they are having a magical power to take care the children and whole family. Usually, pregnancy and care during childbirth were primary focused on the expectant mothers. Much published papers were discussing about mother's experience and how they can cope in pregnancy. But conversely people always neglect the presence of fathers during their partner's pregnancy and after the childbirth. Fathers, in traditional Chinese culture, they are playing a dominant role in a family.

Fathers also recognising as a pillar in the development of the children and fathers are the protectors, the adult who was holding unchallenged power. Their presence and engagement must have a huge impact to their family. Considerable studies have already proven the presence of fathers in pregnancy has great impact to children development. In recent decades, the encouragement of fathers' engagement with children has increased because such kind of involvement were shown improved child cognitive and socio-emotional development. Not only for children, literature review in 2021 reported, shared care with fathers providing greater support to mothers in terms of relieving some of the burden of housework and childcare. Also, may help to improve maternal wellbeing levels and reduce mother's stress. Especially in Hong Kong, no studies have examined the involvement of fathers' during their partner's pregnant also the aspects of the engagement not yet discussed.

Objectives: Aims to shed light and understanding on the experiences of Hong Kong fathers during the period form their partner's pregnancy and childbirth, to bridge the gaps between fathers' involvement and practice in this study. Discover the fathers' experiences in the transitional pathway to fatherhood. To explore and to promote fathers' involvement in their partner's pregnancy and childbirth, an overview of father's involvement should be explored.

The following are the research questions in this study:

1. What are the fathers' perceptions/attitude during pregnancy and after childbirth?
2. How did the fathers' involvement impact the family?
3. What are the mothers' perception/attitude on father's involvement during pregnancy?

Methods: A qualitative study will be conducted. Purposive sampling of a sample of couples will be recruited in antenatal clinic. Following with an interview with interview guides, generate data used to explore the Hong Kong fathers' views and experience during pregnancy. Participants will be interviewed again one month after their partners' delivery. All the data were transcribed and analysed using thematic analysis.

Study implications: Healthcare professionals should identify the fathers' experiences, changes and their needs such as source of informational support, fathers' attitude, relationship with partner and the health outcomes for children. The findings in this study will promote our understanding of the transitional process and aspects of fathers' involvement in Hong Kong during the beginning of their fatherhood life.

Except the process of transitional to fatherhood will be discussed, Family-centred caring (FCC) is the main central idea which should be promoted in the future obstetric care. Support from midwives and nurses appeared to facilitate fathers' involvement during labour. At the moment of birth have already awakening of fatherhood in men, midwives are essential in supporting not only the mothers, but also the fathers. Midwives have the roles to offer them the opportunities to take part and participate in all decision-making process. Therefore, it is absolutely suitable to apply the FCC concept in obstetrics in order to provide the best care to the whole family.

A Quality Improvement Program to Enhance Safety Administration of Expressed Breast Milk (EBM) in Neonatal Unit (NNU)

LP Ho, SM Cheng, YF Lee, MK Wong, AYM Chan

Department of Paediatrics, Prince of Wales Hospital, Hong Kong SAR

Introduction: With the Baby-Friendly Hospital Initiative promote among hospitals, more women are breastfeeding and providing breastmilk to hospitalised infants.

Ensuring safe breastmilk handling and administrating in neonatal unit is a complex process with many potential points for error, of which one of the most serious is administration of the wrong milk to the wrong baby.

Although, our unit has our own guideline specially for proper handling and preparation. We still had few incidents of giving wrong expressed breast milk (EBM) to the wrong babies.

Hence, a new 2D scanning system for milk scanning was employed to promote patient safety and reducing risk of misadministration. A pilot study was initiated from February 2021.

Objectives:

1. To examinate the feasibility of using 2D barcode scanning system for EBM identification
2. To ensure the safe administration of EBM for all infants
3. To enhance staff competence on using the new barcode system

Methodology:

1. To estimate the usages and search for the supply of scanning systems
2. To raise staff awareness on the EBM incidents by providing briefing and training sessions
3. To implement pilot study for testing of efficacy
4. To monitor the efficacy and efficiency of implementation
5. To obtain the evaluation and feedback from staff
6. To compare the number of errors before and after the implementation.
7. To monitor the staff's competence

Results and outcomes: There is no wrong EBM incident in the span of a year (2021).

All staff has shown competence on using the new barcode system.

Rare Thoracic Tumour in a Young Girl: A Call for Multidisciplinary Intervention

YC Chan,[1] ANC Kan,[2] KF To,[3] IYP Wan,[4] BA Rocha,[5] KKF Fung,[6] CSK Lau,[7] YF Cheung,[8,9] CLS Yan,[10] JPW Yau,[10] ECH Fu,[10] CW Luk,[10] MMK Shing,[10] GCF Chan,[9,10] DTL Ku,[10] APY Liu[9,10]

[1]LKS Faculty of Medicine, The University of Hong Kong; [2]Department of Pathology, Hong Kong Children's Hospital; [3]Department of Anatomical and Cellular Pathology, Chinese University of Hong Kong; [4]Department of Surgery, Prince of Wales Hospital, Chinese University of Hong Kong; [5]Division of Cardiothoracic Surgery, Hong Kong Children's Hospital; [6]Department of Radiology, Hong Kong Children's Hospital; [7]Department of Anaesthesiology, Hong Kong Children's Hospital; [8]Division of Paediatric Cardiology, Hong Kong Children's Hospital; [9]Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, The University of Hong Kong; [10]Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Primary thoracic tumours are uncommon in children. The application of next-generation sequencing allows robust disease classification while proactive interventions are required to mitigate potential morbidities associated with mediastinal syndrome.

Purpose and Methods: To describe the clinical course of a child diagnosed with primary thoracic low-grade fibromyxoid sarcoma (LGFMS).

Findings: A previously healthy 9-year old girl presented with six months of dyspnea. Examination showed reduced air entry over the right lung with tracheal deviation contralaterally and facial puffiness. Imaging revealed a giant mass expanding the right thorax with significant compression on the mediastinum. Frozen section on biopsy specimen demonstrated abnormal spindle cells. Chemotherapy was started with the presumed diagnosis of pleuropulmonary blastoma for the life-threatening clinical picture, but with lack of response noted. Subsequently, histologic analysis indicated MUC4 positivity by immunohistochemistry, and *FUS-CREB3L2* fusion was detected by RNA-sequencing, confirmatory of LGFMS. Understanding that surgery is the mainstay, a multidisciplinary team was assembled to devise the peri-operative plan. With pre-operative cannulation of the femoral vessels, embolisation of feeding arteries, and meticulous intra-operative monitoring, near-total resection was achieved without complications. Post-operatively, the patient was pre-emptively maintained on extracorporeal membrane oxygenation due to pulmonary hypertension, and made an uneventful recovery other than

segmental pulmonary embolism managed with enoxaparin. She received intensive pulmonary rehabilitation and demonstrated good exercise tolerance now 7 months from the procedure.

Conclusions: Rare differentials should be considered in children presenting with intra-thoracic tumours. Multidisciplinary management in a tertiary referral center is key to the successful treatment of such high-risk conditions.

Overcoming the Diagnostic and Management Challenges in Metastatic BCOR-Altered Primitive Myxoid Mesenchymal Tumour of Infancy

SH Yu,[1] ANC Kan,[2] KF To,[3] CLS Yan,[4] JPW Yau,[4] ECH Fu,[4] CW Luk,[4] MMK Shing,[4] GCF Chan,[4,5] APY Liu,[4,5] DTL Ku[4]

[1]LKS Faculty of Medicine, The University of Hong Kong; [2]Department of Pathology, Hong Kong Children's Hospital; [3]Department of Anatomical and Cellular Pathology, The Chinese University of Hong Kong; [4]Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital; [5]Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, The University of Hong Kong, Hong Kong SAR

Introduction: Primitive myxoid mesenchymal tumour of infancy (PMMTI) with BCOR alteration is a rare entity newly added to the 2020 WHO Classification of Soft Tissue Tumours.

Purpose and Methods: To illustrate the diagnostic dilemmas and clinical course of the first reported case of PMMTI in Hong Kong.

Findings: A six-month-old girl presented to Hong Kong Children's Hospital with progressive, congenital left dorsal foot swelling. Differential diagnoses considered included infantile haemangioma, overgrowth syndrome and infantile fibrosarcoma. Clinical examination and imaging showed numerous metastatic lesions in the lungs, cervical and inguinal lymph nodes, gluteal muscle, paraspinal region, and subcutaneous tissue. Immunohistochemistry and FISH of left foot biopsy specimen indicated BCOR-positivity with absence of *EWSR1*-rearrangement. *BCOR* internal tandem duplication was then confirmed by RNA-Seq, compatible with diagnosis of PMMTI. Apatinib, temozolomide and irinotecan (AIT) was started with palliative intent. Tumour progressed after 5 cycles, so a course of vincristine, doxorubicin and cyclophosphamide (VDC) was offered, but was complicated by septic shock. VDC was withheld and AIT resumed. Surprisingly, the patient demonstrated partial response and disease stabilisation with continued AIT cycles (21 cycles given to-date). The regimen was well tolerated other than transient hypertension and proteinuria that resolved with apatinib dose reduction. With tumour shrinkage, the girl is now ambulatory and enjoys good quality of life.

Conclusion: Our case highlights the challenges in diagnosing and managing rare paediatric cancers and the critical role of refined histopathologic-molecular workup. Without an established treatment strategy, combination of multikinase inhibitor and chemotherapy resulted in durable disease control.

Evolvement of Childhood OSA Phenotypes After Adenotonsillectomy: Latent Class and Latent Transition Analysis of the Childhood Adenotonsillectomy Trial

Z He, CT Au, AM Li, KC Chan

Department of Paediatrics, The Chinese University of Hong Kong, Hong Kong SAR

Introduction: The severity of obstructive sleep apnoea (OSA) is classically graded by apnoea hypopnoea index (AHI) which is, however, not well correlated with disease complications and treatment outcomes. Identifying phenotypes and evaluating their response to treatment will help to personalise treatment strategy.

Purpose: To classify childhood OSA patients into different phenotypes and to investigate their associations with clinical outcomes and moderating effect on the response to adenotonsillectomy.

Methods: Latent class analysis (LCA) was applied to identify symptom phenotypes using variables from the Paediatric Sleep Questionnaire (PSQ) from the baseline clinical data of the Childhood Adenotonsillectomy Trial (CHAT). The associations of phenotypes with clinical parameters including polysomnography (PSG) measurements, biochemical results, neurobehavioural outcomes and quality of life (QOL) were tested. Differences in phenotypes' response to adenotonsillectomy were further evaluated in two-way ANCOVA (repeated measures). Latent transition analysis (LTA) was performed to assess the phenotype change after adenotonsillectomy.

Findings: Three phenotypes were identified: Phenotype1 (Heaviest "nocturnal and daytime symptoms" burden), Phenotype 2 (Predominantly daytime inattention

symptoms), and Phenotype 3 (Predominantly nocturnal symptoms).

The three phenotypes differed significantly from each other in PSQ and QOL score (all $p<0.05$). Phenotype 3 had significantly lowest cognitive testing score in (Behaviour Rating Inventory of Executive Function) BRIEF, (Child Behaviour Checklist) CBCL, (Conners Rating Scale-Revised) CRSR when compared with Phenotypes 1 and 2 after adjustment for age, gender, BMIs, lgAHI (all $p<0.001$).

After adjustment of age, gender, BMIz, BMIz change, phenotype×time point interaction was significant in PSQ ($P<0.001$), OSA18 ($P<0.001$), CBCL ($P=0.029$) and CRSR ($P=0.008$). However, phenotype × time point × study arm interactions were significant only in lgCRP ($P=0.006$) and PSQ ($P=0.040$).

After adenotonsillectomy, the majority (67%) of Phenotype 1 patients transitioned into Phenotype 2 with the cognitive symptoms unresolved. However, 66% of Phenotype 2 remained as the same phenotype after treatment. 81% of Phenotype 3 patients resolved from the nocturnal symptoms and transitioned into the disease resolution group Phenotype 4.

Conclusions: Using the machine learning algorithms, our study identified three symptom patterns in childhood OSA, with differential associations with clinical outcomes and response to adenotonsillectomy. Phenotype with heaviest symptom burden had higher resistance to improvements. This study provides insights that symptom phenotyping may help to predict treatment outcomes.

Paediatric Fiberoptic Endoscopic Evaluation of Swallowing: Service Review with Clinical Audit on Documentation

OY **Yiu**,[1] KY **Siu**,[2] CSS **Chan**,[1] KY **Choi**,[1] FW **Lo**,[3] R **Fong**[2]

[1]Speech Therapy Unit, Department of Allied Health, Hong Kong Children's Hospital; [2]Department of Otorhinolaryngology, Head and Neck Surgery, Faculty of Medicine, The Chinese University of Hong Kong; [3]Department of Ear, Nose and Throat, Hong Kong Children's Hospital, Hong Kong SAR

Background: Fiberoptic endoscopic evaluation of swallowing (FEES) provides direct visualisation of the anatomy and physiology of swallowing. There is a lack of standardised protocol for FEES performed in children.

Purpose: This study was aimed at systematically evaluating the collaborative ENT and Speech Therapy paediatric FEES performed at Hong Kong Children's Hospital (HKCH) since its inception. The results could guide subsequent service planning and development of paediatric FEES protocol.

Method: A retrospective analysis of 18 FEES performed from October 2019 to August 2021 was done. Demographics of patients, FEES reports and post-FEES progress notes were analysed. They were reviewed independently by two speech therapists using a binary rating system for presence or absence of items based on the protocol used by Cincinnati Children's Hospital Medical Center.

Findings: Half of the patients were below 1-year-old and 61% were female. Oncological disorder was the most common primary diagnosis (50%). The complication rate was 5% (1/18). For FEES documentation, all areas under the four sections of standard protocol were addressed with a reporting rate of 11% to 100%. Some areas were described in details with a reporting rate up to 92% (e.g. location of pharyngeal residue), while some other details were lacking (e.g. amount of aspiration).

Conclusions: Findings indicated FEES is feasible in infants and older children with a low complication rate in HKCH. Clinical documentation covered the essential items including past medical history, anatomy and physiology, secretion management, swallowing deficits. However, inclusion of more detailed documentation varied greatly. The FEES protocol, particularly clinical documentation, should be adhered to in greater details, to facilitate clinical care of patients with dysphagia.

Inferior Endothelial Function Improved Following Optimisation of Anti-Inflammatory Treatment in Asthmatic Children

EC **Wong**,[1] KC **Chan**,[1] CT **Au**,[1] P **Chook**,[2] AM **Li**[1]

[1]Department of Paediatrics; [2]Department of Medicine & Therapeutics, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong SAR

Background: Prior research has explored the potential association between asthma and cardiovascular disease (CVD). However, data has been inconsistent. Confirming the relationship between the two conditions may allow future CVD burden to be lessened following good asthma control.

Purpose: To serially assess endothelial function in physician-diagnosed asthmatic children and to assess the changes in measurements after initiation or up-titration of inhaled corticosteroids (ICS) in those with uncontrolled asthma.

Method: Children aged 6 to 16 years with asthma were recruited to undergo assessment including symptom control (Asthma Control Test (ACT) and Functional Symptom Score (FSS)), spirometry, fractional exhaled nitric oxide (FeNO), and flow-mediated dilation (FMD). FMD is a proven and valid marker of endothelial function. Age-, gender- and body mass index (BMI)-matched non-asthmatic healthy controls was selected for baseline comparison. Asthma subjects were treated with inhaled corticosteroids (ICS) therapy, with repeat measurements carried out at 2 and 4 months from baseline. Linear mixed models were employed to examine effects of ICS therapy across time points.

Findings: Forty subjects were studied, 23 had uncontrolled asthma. Subjects with uncontrolled asthma had significantly lower baseline FMD compared to matched controls (8.0±0.8% vs 8.6±0.7%, p=0.012). Subjects with uncontrolled asthma after treatment showed significantly greater improvements in FMD when compared to the stable asthmatic group after adjustment for age, gender, BMIz (*p* (visit*gp)=0.017). Corresponding improvements were also demonstrated in symptom control, lung function, and FeNO.

Conclusion: Children with uncontrolled asthma had reduced endothelial function compared to matched healthy controls. Significant improvement in endothelial function was demonstrated following better asthma control, suggesting poorly controlled childhood asthma could be an important early-life risk factor for future CVD.

Sleep Duration in Pre-Schoolers is Overestimated by Their Parents

TY **Tsui**, CPK **Chau**, KN **Yu**, AM **Li**, CT **Au**

Department of Paediatrics, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong SAR

Background: Parent-reported sleep diary is the most commonly used tool to collect sleep duration data on pre-schoolers. However, there is a lack of research looking into its accuracy.

Purpose: To validate the sleep duration obtained from 7-day parent-reported sleep diary against objective measurement.

Methods: Data was obtained from a sleep education workshop for pre-schoolers with insufficient sleep (parent-reported sleep duration <25th percentile of a community-based sample). Sleep duration was both reported by parents with a sleep diary and recorded by actigraphy for a week. Data acquisition through actigraphy involved continually wearing a movement (acceleration) sensor on the non-dominant wrist. Levels of agreement between actigraphy and diary recordings were examined by intra-class correlation coefficient (ICC) and Bland-Altman plot.

Findings: Data was successfully obtained from 39 pre-schoolers (13 male, mean age=4.8±0.7) at baseline. The parent-reported average sleep duration was significantly greater than the actigraphy-measured sleep duration (560 min±45 vs. 545 min±49, p<0.001). The ICC between the two measurements was 0.84 (95% CI 0.59-0.93). The agreement between actigraphy-derived and parent-reported sleep of the baseline visit was Mean=15.0 and SD=23.5 min. The 95% CI limits of agreement was -31 to 61 min, with no significant proportional bias.

Conclusion: Despite an acceptable agreement, 7-day parent-reported sleep duration was an over-estimation when compared to the actigraphy-measured sleep duration.

Rehabilitation Outcome of Severe Feeding Disorder in Children with Chronic Illness

QV **Tao**, SL **Lee**

Department of Paediatric and Adolescent Medicine, The Duchess of Kent Children's Hospital at Sandy Bay, Hong Kong SAR

Background: Children with chronic illness frequently have feeding problems as a result of complex interactions between medical, developmental, behavioural and psychosocial factors.

Purpose: This study aims to review the feeding outcome in children with severe feeding disorder due to chronic illness after participating in a multidisciplinary feeding program.

Methods: In this retrospective study, we reviewed the medical records of children admitted to a paediatric rehabilitation centre for intensive feeding program between January 2017 and December 2018, with follow-up upto July 2021. Patient demographics, anthropometric parameters, medical background, swallowing assessment and tube feeding duration were recorded.

Findings: Among the 18 children (age 2m to 13yr) who completed the training program, ten (55%) successfully achieved full oral feeding. Four (22%) weaned off feeding tube at the end of program. Six (33%) weaned off feeding tube when receiving follow-up out-patient service. The

median tube feeding duration was 18 months (range 8 to 50 months) in children who achieved full oral feeding. Among them, seven children (70%) developed behavioural feeding problems, a complication associated with prolonged tube feeding. Six out of fourteen (43%) children with medical complexity achieved full oral feeding after training. Global developmental delay was common (94%) in our cohort, indicating multidisciplinary neurodevelopmental training in addition to feeding program.

Conclusion: In children with severe feeding disorder due to chronic illness, oral feeding rehabilitation took time and required multidisciplinary approach. Besides medical follow-ups, the patients and their families often required resources to help them face challenges in developmental, behavioural and psychosocial aspects.

Back-To-Back Comparison of Diagnostic Efficacy Between Genome Sequencing and Exome Sequencing Reanalysis in Primary Ciliary Dyskinesia Cohort

JFT Chau,[1] MHC Yu,[1] JLF Fung,[1] CCY Mak,[1] M Lee,[1] MHY Tsang,[1] KS Yeung,[1] SL Lee,[2] BHY Chung[1*]

[1]Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, The University of Hong Kong; [2]Department of Paediatrics and Adolescent Medicine, Duchess of Kent Children's Hospital, Hong Kong SAR

*Corresponding author

Background: Primary ciliary dyskinesia (PCD) is a heterogenous genetic disorder with more than 44 genes correlated. With the rapid improvement of next generation sequencing, exome sequencing is frequently utilised in the clinical setting for molecular diagnosis. However, there is still an ongoing debate on whether to proceed with genome sequencing (GS) or perform exome sequencing reanalysis (rES) if the initial results return negative. By using a back-to-back double-blinded comparison, this study aims to evaluate between the two methodologies. This comparison serves as the first to inform clinicians and clinical geneticist on the optimal next action after a negative exome sequencing result.

Methods: In this study from 2015 to 2020, 55 participants (29 males and 26 females) suspected of PCD were recruited into this study and exome sequencing was performed. Negative or inconclusive exome sequencing results were considered for GS due to high genotype-phenotype correlation. GS was then performed on 30 (13 males and 17 females) inconclusive exome sequencing results. In GS, 50% concordant CNVs calls from ERDS and CNVnator were analysed in this study. Two teams of genome analysts and bioinformaticians were randomly allocated to GS or rES and were blinded to the other team's analysis. The time for bioinformatics, analysis, and discussion were also recorded for evaluation.

Findings: Exome sequencing revealed 5 positive cases in the initial batch of exome sequencing. The positive case included mutations in *RSPH4A*, *CCDC40*, *DNAH11*, and *CFTR* (2 cases were correlated to cystic fibrosis). GS and rES, in this cohort, achieved one new diagnosis and identified eight VUS results with potential clinical relevance. The new diagnosis in *DNAH11* was made due to an update in medical literature which upgraded a VUS to likely pathogenic, thereby confirming the diagnosis. This variant was identified in both GS and rES. GS was also able to detect an additional VUS result due to low coverage in rES within exon 3 of the *DNAAF3* gene. The patient's functional result matches previously reported *DNAAF3* static ciliary movement.

Conclusion: Beyond deep intronics changes, CNVs, and SVs, GS had an additional advantage of uniform coverage over the exons. Here we have investigated a back-to-back comparison in diagnostic efficacy between GS and rES. Further investigations are needed to explore deep intronic mutations and structural variations in cases with one VUS mutation found.

Acknowledgement: This study was supported by the Society for the Relief of Disabled Children, the Health and Medical Research Fund (HMRF), Li Ka Shing Donation Account: Enhanced New Staff Start-up Packages, and the Edward and Yolanda Wong Fund.

Establishment of Hong Kong Neuromuscular Disorder Patient Registry

MKL **Yu**,[1] WHS **Wong**,[1,2] Y **Gao**,[3] JS **Rosa Duque**,[1,2] GCF **Chan**,[1,2] SHS **Chan**[1,2]

[1]Department of Paediatrics and Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong; [2]Department of Paediatrics and Adolescent Medicine, Queen Mary Hospital; [3]Division of Neurology, Department of Medicine, Queen Mary Hospital, The University of Hong Kong, Hong Kong SAR

Introduction: Neuromuscular disorders (NMDs) are a group of diseases affecting the peripheral nervous system (1). Many NMDs cause disability or even premature death (2). We aim to design and establish a robust NMD patient registry in Hong Kong.

Methods: By modelling international NMD patient registries, we designed patient-professional reported questionnaires to collect the demographic, clinical characteristics, genetic details, family history, investigation findings and specific treatment of NMD patients. Patients were recruited through Hong Kong West Cluster (DKCH, QMH) and Kowloon Central Cluster (HKCH). We also developed self-registration online platform. $p<0.05$ was considered statistically significant.

Findings: Since June 2019, 125 NMD patients have been enrolled in the registry with 12 participants registered online. The registry recruited 13 types of NMDs, including spinal muscular atrophy (SMA) (n=31), Duchenne muscular dystrophy (DMD) (n=19) and congenital myopathy (n=18). The age range was 7 months to 63 years old. 65.6% of those enrolled were children (<18 years old). 63.2% were male. 64.8% of the patients had genetic diagnosis. The registry has contributed to two studies. The first one is a prospective study of clinical efficiency of Nusinersen in SMA patients (n=22). 14/16 SMA patients showed improvement in at least one of motor performance (CHOP intend/RULM/HINE/HFMSE) and health-related quality of life after 1st year of treatment. The second study is the reactogenicity and immunogenicity study of the COVID-19 vaccine in DMD patients (n=4). Data will be available in October.

Conclusion: Hong Kong Patient registry has contributed to ongoing and new research study to optimise medical care.

Natural History of Acute Kidney Injury Among Critically Ill Children

WF **Hui**, KL **Hon**

Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Acute kidney injury (AKI) among critically ill patients contributes to both morbidity and mortality. Local data on progression of AKI among critically ill children is lacking.

Purpose: We described the epidemiology of AKI among children admitted to the paediatric intensive care unit (PICU) of the Hong Kong Children's Hospital. This is the interim analysis of the prospective cohort study (E-AKI-DRUG) conducted in our PICU.

Methods: All children aged 1 month to 18 years old would be enrolled. Exclusion criteria included those with pre-existing chronic kidney disease, impaired renal function for ≥3 months, immediate post-renal transplant and short PICU stay of ≤1 days. Children with no urinary catheter would be excluded from urine calculation. AKI would be defined using the KDIGO criteria. The data of initial four months would be presented.

Findings: We identified 62 children with 63 episodes of admission for this analysis. Male accounted for 59% of the admissions and the median (interquartile range) age was 6.1 (6.6) years old. The overall incidence of AKI during PICU stay was 48.4% (Stage 1: 20.3%; Stage 2: 12.5%; Stage 3: 15.6%). 33.3% of children already developed AKI on PICU admission and 53.3% of children attained the highest stage of AKI on day 1 of admission. The median duration of AKI during PICU stay was 1 (3) day. Most children with AKI were non-oliguric with their urine output during PICU stay being 3.9 (2.9) ml/kg/hour. Among children with AKI, only 32.3% of them had their serum creatinine level (SCr) returning to baseline level. The duration from peak SCr to lowest SCr was significantly longer among those with higher stage of AKI (p=0.002). CRRT was required among 6.3% of children. Upon PICU discharge, AKI was not yet resolved among 38.7% of patients (Stage 1: 12.9%; Stage 2: 16.1%; Stage 3: 9.7%) and 3.1% of the patients remained dialysis-dependent. The staging (p<0.001) and the duration of AKI (p=0.018) were both associated with worse stage AKI at PICU discharge. Overall PICU mortality was 4.7% and AKI was associated with longer PICU stay (5 vs 3 days, p=0.003) and higher mortality (12.9% vs 0%. p=0.05).

Conclusion: AKI was common among critically ill children and most of them acquired the condition on 1st day of PICU admission. AKI was not resolved in a significant proportion of children upon PICU discharge, and a higher AKI stage and longer AKI duration were associated with worse renal outcome at PICU discharge.

Acute Kidney Injury and Electrolytes Disturbances Among Critically Ill Children with Malignancy

WF **Hui**,[1] VPY **Chan**,[2] KL **Hon**,[1] F **Poon**[2]

[1]Department of Paediatrics and Adolescent Medicine; [2]Department of Pharmacy, Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Acute kidney injury (AKI) and electrolyte disturbances are common among critically ill children and contributes to both morbidity and mortality. Children with oncological diagnosis is at risk of developing both conditions. We described the epidemiology of AKI and electrolytes disturbances among children with malignancy admitted to the paediatric intensive care unit (PICU) of the Hong Kong Children's Hospital.

Methods: This is the interim analysis of the prospective study on the epidemiology of AKI and electrolytes disturbances (E-AKI-DRUG) conducted in our PICU. All children aged 1 month to 18 years old would be enrolled, except those with pre-existing chronic kidney disease, impaired renal function for ≥3 months, immediate post-renal transplant and short PICU stay of ≤1 days. Children with no indwelling urinary catheter would be excluded for urine calculation. AKI would be defined using the KDIGO criteria and serum electrolytes profiles on sodium, potassium, calcium, phosphate and magnesium were reviewed. The data of initial four months would be presented.

Findings: Altogether 63 episodes of admission were enrolled. 59% were male and median (interquartile range) age was 6.1 (6.6) years old. 49.2% of patients were oncology patients. The overall incidence of AKI during PICU stay was 48.4% (Stage 1: 20.3%; Stage 2: 12.5%; Stage 3: 15.6%). Children with malignancy had a significantly higher PIM3 score (p=0.014), more use of mechanical ventilation (p=0.041) and higher number (p<0.01) and doses (p=0.023) of nephrotoxic medication exposure than those without malignancy. Oncology patients had a significantly higher incidence of AKI compared to non-oncological patients (62.5% vs 34.4%, p=0.024). Hypophosphataemia, hypocalcaemia and hypokalaemia were the most common electrolytes disturbances among oncology patients (incidence of 87.5%, 77.4% and 56.3% respectively). There was significantly more hyponatraemia among oncological patients (43.8% vs 9.7%, p<0.01). Tubular dysfunction was common among children with electrolytes disturbance and oncological patients had significantly more urinary phosphate wasting than those without malignancy (66.7% vs 33.3%, p=0.046).

Conclusion: Children with malignancy admitted to the PICU had a higher risk of developing AKI and electrolytes disturbances. Tubular dysfunction, especially urinary phosphate wasting, was commonly observed among children with malignancy having electrolytes disturbances.

Gene Burden Analysis Identified Candidate Genes Enriched in Exomes of Non-Syndromic Conotruncal Cardiac Defects Patients in Hong Kong

MMC **Chui**, CCY **Mak**, JFT **Chau**, MHC **Yu**, PC **Chow**, KS **Lun**, TC **Yung**, BHY **Chung**

Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, The University of Hong Kong, Hong Kong SAR

Background: Conotruncal cardiac defect (CTD) is a group of congenital heart diseases with an estimated prevalence of 11.6 per 10,000 live births worldwide, and is potentially more prevalent in Chinese. Nonetheless, no genetic cause can be identified for most non-syndromic CTD. Hence this study aimed to utilise whole exome sequencing (WES) to identify candidate genes associated with non-syndromic CTD in local population.

Methods: Patients diagnosed with CTD and negative for 22q11.2 Deletion Syndrome QF-PCR were recruited from the cardiac clinic at Queen Mary Hospital. Gene burden analysis was performed on WES data based on a high-confidence congenital heart disease gene list consisting of 132 genes. An excess of rare (allele frequency <0.01% in gnomAD) protein-altering variants compared to two ethnically matched population sets, 853 local individuals without cardiac diseases and 9,197 gnomAD East Asian controls, was identified by Fisher's exact test. A systematic review of relevant literature was performed for each prioritised candidate gene to support their roles in outflow tract development.

Findings: In a cohort of 265 predominantly Chinese non-syndromic CTD patients, nine candidate genes were

prioritised by gene burden analysis. The excess of rare protein-altering variants strengthened the gene-disease association of three established genes (*GATA6, NOTCH1, TBX1*) for tetralogy of Fallot. This study also provided additional evidence for the potential roles of other prioritised genes (*ANKRD11, ARID1A, MYH6, SOS1, STRA6, TRAF7*) in the complex aetiology of CTD. Four of these genes were previously reported to be related to syndromes with extracardiac features but not identified as genetic causes of isolated CTD.

Conclusion: Our results substantiated the important genotype-phenotype associations in the largest Chinese cohort of CTD in Hong Kong. Candidate genes associated with CTD related syndromes were enriched in this non-syndromic cohort, providing novel insight into the complex genetic architecture of CTD.

Acknowledgements: This study was supported by grants from the Society for the Relief of Disabled Children and the Children's Heart Foundation of Hong Kong.

Establishment of a Paediatric Palliative Care (PPC) Team in Department of Paediatrics

SP Law

Prince of Wales Hospital, Hong Kong SAR

Background: Palliative care should be commenced once illness is diagnosed, rather than at decline, and it should be continued. With the increasing number of children with complex chronic conditions or sudden complex illnesses, as well as the needs of family-centered care, Paediatric Palliative care (PPC) is crucial and should not be delayed. In HK, the PPC service provided by Hong Kong Children's Hospital and NGOs were not always 24/7 available, and therefore it was necessary to form a local PPC team to readily intervene for those patients and families in need.

Purpose: To establish a team to provide PPC service.

Methods: A PPC workgroup was formed in 2020. PPC policy and documentation were standardised to improve workflow and clinical handover. Collaboration among professionals across different sectors and NGOs was facilitated for comprehensive intervention. Patients and families were supported for their physical, psychological, social and spiritual problems. PPC trainings were provided for nurses and doctors to raise awareness and enhance their skills and knowledge.

Findings: Total 36 referrals were made by physicians, the families were interviewed by PPC team within 0 to 3 days. The families were being referred to the professionals across different sectors and NGOs for support according to their problems and needs. End-of-life care and bereavement care were provided to the families of all the deceased patients.

Conclusion: A systematic PPC service could not only increase the quality of care, but also enhance family-centered care, and promote quality of life for the patients and their families.

Eczema in Hong Kong: Prevalence Trends and its Association with Breastfeeding

KW Kwok, OC Chun

The Chinese University of Hong Kong, Hong Kong SAR

Introduction: Eczema is a common childhood allergic disease which significantly affects patients' quality of life.

Purpose: To review the trend of childhood eczema in Hong Kong and investigate its association with breastfeeding.

Methods: Parents were recruited from kindergartens in Hong Kong from late 2020 to mid-2021, to complete a standardised questionnaire adapted from the ISAAC. The local prevalence of childhood eczema was retrieved from two territory-wide cross-sectional studies conducted in 2001 and 2009. The prevalence of childhood eczema was expressed as a proportion with a 95% confidence interval (CI). The association between child eczema and the aforementioned factors was evaluated using univariate and multivariate analysis.

Findings: The prevalence of lifetime eczema and current eczema increased significantly from 31.9% in 2009 to 37.2% (95% CI: 0.35-0.40, $p<0.001$) in 2020 and from 4.5% in 2001 to 11.5% (95% CI: 0.10-0.13, $p<0.001$) in 2020 respectively. Childhood eczema was positively associated with a family history of eczema (OR=2.71, $p<0.001$) and breastfeeding (OR=1.41, $p=0.006$). After controlling for family history, the odds of having eczema increased by 1.05 times (95%CI 1.01-1.07, $p=0.002$) per 6-month increase in breastfeeding time, and 1.05 times (95%CI 1.02-1.08, $p=0.002$) per 2-month increase in exclusive breastfeeding time. Yet, children with an early onset of eczema were significantly more likely to be breastfed ($p=0.001$) and exclusively breastfed ($p=0.002$) for a longer duration.

Conclusion: The prevalence of childhood eczema has significantly increased over the decade. While genetic factor is the key determinant of childhood eczema, breastfeeding is a risk factor for eczema after controlling for family history in this study.

Noise Reduction Program in Neonatal Intensive Care Unit (NICU)

LP Ho, PF Ho, WY Ip, HW Leung, KY Yeung, SM Cheng
Department of Paediatrics, Prince of Wales Hospital, Hong Kong SAR

Introduction: Infants in NICU are constantly exposed to ambient noise that often exceeds recommended levels. Premature Infants are particularly vulnerable to elevated noise levels due to their immature auditory pathway. Many studies showed that such excessive noise will lead to many adverse health consequences. We noted that noise level in our NICU was ranged from 52 to 79 dB that often exceeded the AAP recommended (45dB) levels. Although some loud noise in the NICU is unavoidable, strategies were implemented to decrease the noise levels in our busy NICU

Objectives:

1. To explore the sources of excessive loud noise in our NICU
2. To implement the strategies to alleviate noise sources, and
3. To enhance staff awareness of maintaining a quiet environment

Methodology:

1. To perform literature review on the evidence of noise source in NICU
2. To study the current noise sources and staffs' perception on sources of noise in local NICU
3. To raise staff awareness on the impact of loud noise by providing briefing and training sessions
4. To implement strategies for noise reduction
5. To monitor the noise level in NICU
6. To compare the noise level before and after the program.
7. To monitor the staff's behavioural change after implementation of the noise reduction program

Results and outcomes: The mean ambient noise level was lowered within NICU by 9% from baseline measurements in the span of a year. There was also decreasing in individual items from 0.3 db to maximum 19.7 db after implementation of difference strategies. Staff's behaviour was continuous monitoring, and the program was sustaining.

Challenges in Nutritional Support for Children with Brain Tumour: An Adaptive Solution Against All Odds

GY Wong,[1] HHY Lau,[2] YCL Leung,[2] JPW Yau,[1]
NSY Chao,[2] MWY Leung[2]
[1]Department of Paediatrics & Adolescent Medicine; [2]Department of Surgery,
Hong Kong Children's Hospital, Hong Kong SAR

Introduction: Feeding disorders are commonly observed in paediatric neuro-oncology patients. The causes are multifactorial involving neurological dysfunction, treatment related emesis, mucositis and behavioral changes. Potential complications of malnutrition including electrolyte disturbances, growth deprivation, increased risks for infection and poor quality of life are often overlooked.

Method: Case-report: A 5-year-old boy initially presented with obstructive hydrocephalus. Imaging and pathology confirmed the diagnosis of classic medulloblastoma group-3 with extensive leptomeningeal metastasis. Tumour excision was performed, followed by multiple ventriculoperitoneal-shunt revisions and chemoradiotherapy. Poor feeding and weight-loss persisted despite strategic nutritional consultations because of continued vomiting with intolerance to nasogastric/ nasojejunal tube placement. Prolonged rescue total-parenteral-nutrition was required to regain weight from <3rd to 25th percentile. Feeding-gastrostomy was considered while isotope-milk-scan demonstrated significant delayed gastric-emptying. Primary transgastric-jejunal (TGJ) tube insertion proceeded eventually upon optimal recovery of blood-counts after cycle-2 maintenance-chemotherapy plus G-CSF therapy. Intraoperatively, ventriculoperitoneal-shunt in-situ at gastric-position was localised and safe-guarded fluoroscopically and laparoscopically prior to siting of gastrostomy in close proximity; low-profile-TGJ tube was inserted 'Seldinger' fashion uneventfully. With expectant suboptimal wound healing postoperatively, extended antibiotic-prophylaxis and proton-pump-inhibition was adopted to minimise shunt-contamination risks.

Outcomes: Recovery was uneventful with early escalation to full jejunal-feeding. Adaptive/alternating gastric and jejunal-feeding regimen correlating subsequent chemotherapy-cycles was adopted and well-tolerated at home, regaining weight from 25th to >50th percentile.

Conclusion: Primary TGJ placement is novel in our local experience. All due technical challenges and oncological/operation-risks must be considered in

transdisciplinary setting. It facilitates adaptive enteric feeding regimen when appropriate, a promising strategic option amongst paediatric neuro-oncology patients acquiring difficult feeding-disorders.

Urinary Tract Infection in Infants Younger Than 2 Months – Which Imaging Approach to Adopt?

SW **Wong**,[1] SN **Wong**[2]

[1]Tuen Mun Hospital/Hong Kong Children's Hospital; [2]Department of Paediatrics and Adolescent Medicine, Tuen Mun Hospital, Hong Kong SAR

Aims: A well-established imaging guideline for urinary tract infection (UTI) in infants younger than 2 months old is lacking. We analysed the characteristics of this group of patients to look into the possibility of adopting a more selective imaging approach for investigation.

Methods and Material: This is a post-hoc analysis using data of the patient cohort collected between 2005 and 2006 for the study by Wong et al in 2010.

Results: 170 patients were selected and reviewed. Eleven patients with significant urological abnormalities were identified. They had at least one of the following characteristics: (1) atypical features, (2) abnormal urinary ultrasound (USG) report and (3) UTI recurrence within the follow-up period.

Conclusions: Following first occurrence of UTI, infants younger than 2 months old presenting with atypical features, abnormal USG of the urinary system, or recurrence of UTI may warrant more extensive investigations with micturating cystourethrogram (MCUG) and dimercaptosuccinic acid (DMSA) scans. For those with typical features at presentation, USG of the urinary system alone may be adequate as basic screening, unless UTI recurs. Further studies are required to establish a specific guideline for this group of patients.

Challenges in the Management of an Infant with Matthew Wood Syndrome Having Pulmonary Hypoplasia and Visual Impairment: A Case Report

KL **Wong**, KL **Siu**

Department of Paediatrics, Queen Elizabeth Hospital, Hong Kong SAR

Background: Matthew Wood syndrome (MWS) is a rare entity in which the two main characteristics include anophthalmia and pulmonary hypoplasia. Other problems such as diaphragmatic eventration, duodenal stenosis, pancreatic malformations, intellectual disability, cardiovascular abnormalities, and intrauterine growth retardation have also been reported.

Purpose: This is a case report discussing the challenges encountered in the long-term care of a patient with MWS.

Findings: We report a five-year-old boy who had MWS diagnosed in the neonatal period presenting with bilateral microphthalmia, bilateral pulmonary hypoplasia, diaphragmatic eventuation, and congenital cardiac defects. Misalignment between the light-dark cycle and the endogenous circadian timing causes circadian rhythm sleep disorder (CRSD) – nonentrained type. With nonentrained type CRSD, MWS patients might be somnolent during the day while insomnia is experienced at night. Sleep disturbances have a great impact on the quality of life of this group of patients and limit their opportunity of training in the daytime.

On the ground that they have visual impairment, cognitive delay and craniofacial dysmorphism, numerous difficulties are encountered during the initiation of non-invasive ventilation. We did not target a perfectly normal blood gas reading as the treatment goal, rather a balance on optimal ventilatory support against safety and patient's comfort are of a pivotal importance.

Conclusion: Main clinical features of MWS include anophthalmia/microphthalmia and pulmonary hypoplasia or agenesis. CRSD in this group of patients and constraint on optimising ventilatory support were important issues in their long-term care.