

## MCQs

### Instruction:

1. Please use pencil to shade the box for the best and correct answer (only one answer for each question).
2. Send back the answer sheet (see loose leaf page) to the Hong Kong College of Paediatricians. One point will be awarded to each article if  $\geq 3$  of the 5 answers are correct. The total score of the 4 articles will be 4 CME points.

### (A) Genetic Analysis of Wilson Disease in South China: Hotspots and One Novel Mutation in *ATP7B*

1. The mutations in *ATP7B* would commonly cause harms to the organs except?
  - a. Brain
  - b. Liver
  - c. Bone
  - d. Cornea
  - e. Kidney
2. Are the mutations or polymorphisms on *ATP7B* clustered or dispersed?
  - a. Dispersed
  - b. Clustered within hotspot exons
  - c. The same with that of the Europeans'
  - d. Not certain
  - e. Not clustered in European
3. What's the major chemical marker for diagnosing Wilson disease in clinical trial now?
  - a. The serum concentration of Copper
  - b. Alanine Transaminase
  - c. Aspartate Transaminase
  - d. Ceruloplasmin
  - e. None
4. Which of the following is not the symptom of Wilson disease?
  - a. Liver cirrhosis
  - b. Cornea ring
  - c. Dyskinesia
  - d. Cognition problem
  - e. Shortness of breath
5. Which of the following about Wilson disease is false?
  - a. It's an autosomal recessive inherited disorder
  - b. Directly caused by accumulation of CU
  - c. Could be diagnosed only based on specific symptom
  - d. Earlier diagnose is much more meaningful
  - e. Genetic analysis is indispensable for diagnosis

### (B) Comprehensive Mutation Analysis of the RAS/RAF/MEK/ERK Pathway in Paediatric Leukaemia and Significant Inferences

1. In which percentage of all cancers types is the RAS pathway mutated?
  - a. 15%
  - b. 45%
  - c. 30%
  - d. 10%
  - e. 5%
2. In which of the following options are the most mutated RAS codons given correctly?
  - a. Codons 12,13
  - b. Codons 25, 23
  - c. Codons 13
  - d. Codons 12,65
  - e. Codons 12,13,61
3. ERK1/2 is involved in vital events for cell physiology such as?
  - a. cell proliferation, differentiation, transcriptional regulation, and cellular development
  - b. signal transduction
  - c. programmed cell death and cell differentiation
  - d. histone acetylation
  - e. haemostasis
4. MEK1 and MEK2 are dual-specificity kinases that activate ERK1 and ERK2 by phosphorylating them at conserved?
  - a. arginine and tyrosine residues
  - b. threonine and tyrosine residues
  - c. cysteine residue
  - d. two tryptophan residues
  - e. arginine and threonine residues
5. In which cancer types are ERK mutations most common?
  - a. brain cancer
  - b. bladder and stomach cancers
  - c. leukaemia and lymphoma
  - d. cervix, endometrium, and skin cancers
  - e. lung and skin cancers

**(C) Analysis of Hearing Loss-Associated Gene Mutations Using MALDI-TOF Mass Spectrometry in Neonates from Shaoxing, China**

1. What is the cause of deafness?
  - a. Genetic factors
  - b. Drug-induced
  - c. Environmental factors
  - d. Trauma and noise
  - e. All of the above
2. The newborn hearing screening technology used in China mainly include?
  - a. DPOAE
  - b. AABR
  - c. Acoustic immittance
  - d. Pure tone audiometry
  - e. A and B
3. Four common deafness- susceptibility genes in Chinese population?
  - a. 12S rRNA
  - b. GJB2
  - c. GJB3
  - d. SLC26A4
  - e. All of the above
4. What technology is used for detecting hearing loss-associated genes in this paper?
  - a. PCR-RFLP
  - b. HRM
  - c. NGS
  - d. MALDI-TOF mass spectrometry
  - e. Sanger sequencing
5. What is the sensitivity and specificity of MALDI-TOF-MS for analysing twenty hotspot hearing-associated mutations of four common hearing loss-susceptibility genes?
  - a. 50%
  - b. 75%
  - c. >99.9%
  - d. 90%
  - e. ≤50%

**(D) Clinic Analysis on 4 Infantile Citrin Deficiency Cases**

1. What biochemical abnormalities can be caused by citrin protein deficiency?
  - a. Abnormal liver function
  - b. Hyperlactacidemia
  - c. Hyperammonemia
  - d. Hypoglycemia
  - e. All of the above
2. What is the best diet for neonatal intrahepatic cholestasis (NICCD) patients?
  - a. Lactose-free and MCT-enriched diet
  - b. Carbohydrate rich diet
  - c. High protein diet
  - d. High fat diet
  - e. Low fat diet
3. What vitamins do NICCD patients need to supplement?
  - a. Vitamin A
  - b. Vitamin D
  - c. Vitamin E
  - d. Vitamin K
  - e. All of the above
4. Which amino acid transport disorder is affected by insufficient function of citrin protein?
  - a. Citrulline
  - b. Aspartic acid
  - c. Tyrosine
  - d. Phenylalanine
  - e. Leucine
5. Which amino acids are elevated in citrin protein deficiency patients?
  - a. Citrulline
  - b. Methionine
  - c. Threonine
  - d. Arginine
  - e. All of the above

***Answers of January issue 2021***

(A) 1. a; 2. e; 3. e; 4. e; 5. c

(B) 1. d; 2. c; 3. d; 4. c; 5. e

(C) 1. c; 2. b; 3. b; 4. e; 5. e

(D) 1. a; 2. e; 3. e; 4. a; 5. b