
Clinical Quiz

What is the Diagnosis?

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Our proband was referred to us at 12 years of age. He was born at full term by lower segment caesarian section due to cephalopelvic disproportion and has a birth weight of 3.9 kg. Apart from the enlarged head, he was asymptomatic and his perinatal history was uneventful. Due to the large head size, computed tomography was performed at 2 years and 3 months old and our proband was incidentally found to have a medulloblastoma. The tumour was surgically removed and gross total resection was achieved after 2 operations. Only chemotherapy without radiotherapy was given. Malocclusion was accidentally discovered and panoramic X-ray in 2013 showed multiple mandibular odontogenic keratocyst

as well as calcifications at anterior cerebral falx and tentorium.

Physical examination was carried out during the consultation. The head circumference of our proband was 2 cm above the 97th centile line on the growth curve and his height and weight were lying on the mean for his age. He was noticed to have palm pits, scanty naevi over skin, pectus excavatum and scoliosis which concave to the right. The proband currently attends normal main stream school, but is noticed to have dysarticulation, mild dysmetria and dysdiadokokinesia and he cannot walk from heel to toe. Clinical photographs taken at 12 years of age and X-rays can be seen in Figures 1 & 2 respectively.

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Figure 1 Clinical photographs of our proband at 12 years of age (Palm pits are circled in the top middle photograph). (Parental consent obtained for the clinical photo)

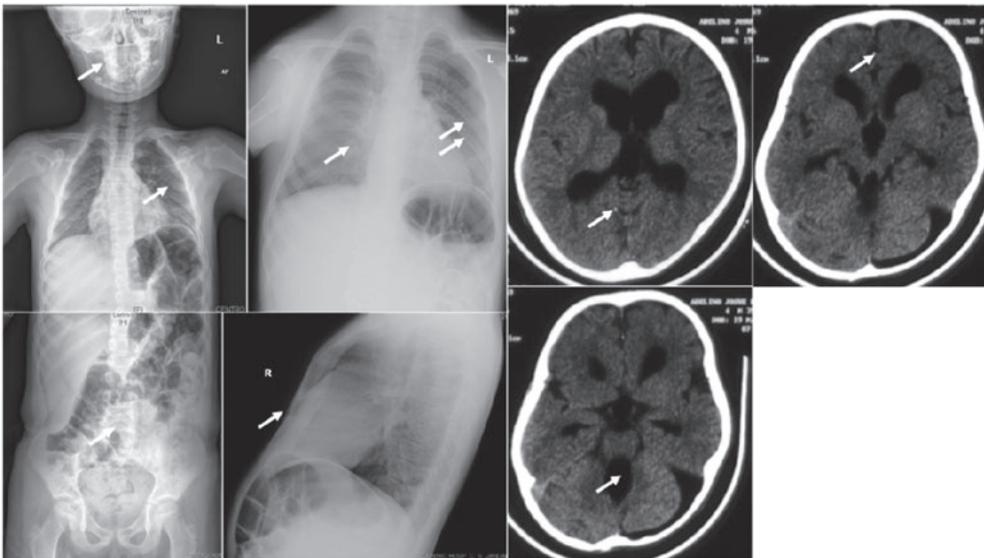


Figure 2 Selected radiographs of our proband. XR Thoracic Spine (AP) included part of mandible, demonstrating well-defined lytic lesion in right mandible, in keeping with keratocystic odontogenic tumour. XR Lumbrosacral Spine (AP) showed spina bifida occulta at L5 and S1. CXR (PA) revealed bifid left 3rd rib and suggested pectus excavatum deformity, which was confirmed in CXR (lateral). CT Brain showed calcifications along falx cerebri and tentorium. Volume loss in cerebellum and ballooning of 4th ventricle are likely due to previous surgery for medulloblastoma.

Answer to "Clinical Quiz" on Pages 128-132

N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.