Hydrocephalus in Hajdu-Cheney syndrome

Hajdu-Cheney syndrome, also known as type VI idiopathic osteolysis or acro-osteolysis, is a rare autosomal dominant disorder with variable expressivity, characterised by a distinctive facies, abnormal skull shape, premature loss of teeth, osteolysis of the terminal phalanges, and short stature. We would like to draw attention to the complication of basal invagination and hydrocephalus in a child with this condition. To our knowledge, this finding has hitherto only been recorded in radiological,[1,2] neuroradiological,[3] and surgical[4] publications in symptomatic adults.

The male proband was the second child of healthy, unrelated parents. The pregnancy was normal with delivery by cesarean section because of fetal distress. Birth weight was 3610 g. Features noted at birth included generalised hyptonia, webbed neck, synophrys, low set ears, high arched palate, proximal limb shortening, right radioulnar dislocation, broad and proximally inserted thumbs, bilateral transverse palmar creases, broad toes, bilateral talipes equinovarus, right metatarsus adductus, short sternum, pectus carinatum, patent ductus arteriosus, small ventricular septal defect, umbilical and bilateral inguinal herniae, and cryptorchidism.

As a young child there were recurrent respiratory infections, asthma, and serious otitis media and he was found to have bilateral conductive hearing loss.

Additional features noted at 7 years include mild mental retardation, occipital prominence, coarse dark eyebrows, prominent coarse eyelashes, upward slanting palpebral fissures, puffy upper eyelids, telecanthus, poorly developed supraorbital margins, posteriorly rotated ears, featureless philtrum, delayed dentition, mild micrognathia, and poorly developed subcutaneous tissues of the face. Sweat pores were prominent in the axillae, groins, and over the anterior and lateral neck skin. The toe nails were dystrophic and there was koilonychia of the index fingers of both hands. Surgical scars were hypertrophic. He had mild persistent hepatosplenomegaly.

The following investigations gave normal results: G band karyotype, thyroid function tests, urinary mucopolysaccharides, urinary organic and amino acids, and lysosomal enzyme activity in peripheral blood leucocytes and cultured fibroblasts.

Radiographs showed abnormal tubulation of all long bones with metaphyseal expansion, longitudinal striations, and sclerosis of the mid-tibial diaphyses, distal tibiae and fibulae, metatarsal cortices, and posterior tali. There were submetaphyseal lucencies of the proximal humeri, dislocation and subluxation of the right radial head, broad corticosteric ribs, mild thoracic kyphosis, evidence of old metatarsal fractures with pseudarthrosis at a previous fracture site, osteolysis of several terminal phalanges of the fingers and toes (fig 1), and metatarsus varus. The lumbar spine showed a ‘bone within bone’ appearance with increased density of vertebral body end plates and prominent inferior scalloping of lumbar vertebral bodies.

At 10 years of age (fig 2), skull radiograph (fig 3) showed generalised thickening of the cranial vault with occipital prominence, multiple wormian bones, enlarged infantile shaped sella turcica, and sclerosis of the skull base. There was osteoporosis of the vertebral bodies. CT scan of the head, performed to delineate middle ear anatomy, showed hydrocephalus, enlarged internal auditory canals, a horizontal clivus, prominent pituitary fossa, distorted foramen magnum, and marked platybasia associated with basal invagination. He was found to have optic disc swelling on funduscopy; visual acuity was 6/6 and 6/9 in the left and right eyes respectively. He was asymptomatic and the remainder of the neurological examination was unremarkable. MRI (fig 4) showed the following features: marked hydrocephalus with Arnold-Chiari malformation and obstruction to cerebrosinal fluid flow at the level of the foramen magnum, an expanded pituitary fossa, distortion of the optic nerves and infundibular stalk, inversion of the clivus.

We present this case to alert clinicians to the potential complication of basal invagination and hydrocephalus in children with Hajdu-Cheney syndrome. This case shows that even in asymptomatic children in whom neurological examination is normal, severe basal invagination and hydrocephalus may occur. These complications should be looked for in children with Hajdu-Cheney syndrome.