Brainstem and cerebellar hypoplasia associated with osteogenesis imperfecta type-5

Sir,

Osteogenesis imperfecta type V is characterized by multiple bone fractures with hyperplastic callus formation in patients with white sclerae and usually negative family history of the disorder. Neurological complications associated with Osteogenesis imperfecta (OI) include basilar invagination, brainstem compression, hydrocephalus and syringohydromyelia. There is only one case report of OI type 4 associated with cerebellar hypoplasia in a Chinese infant. The proposed mechanism for cerebellar hypoplasia is in utero vascular compromise by compression of posterior circulation due to associated craniovertebral anomalies. Herein we report a case of brain stem and cerebellar hypoplasia associated with osteogenesis imperfecta type 5, which may support the hypothesis of vascular compromise as the cause of brain stem cerebellar hypoplasia in OI.

A five-month-old male child born of second-degree consanguinous marriage was admitted with complaints of developmental delay and fractures of both arms. Antenatal, natal, postnatal periods were uneventful. The mother noticed visual impairment and deafness and child was sleeping with eyes half open. On examination, the child had microcephaly, flat occiput and dysmorphic facies (microphthalmia, low set ears and micrognathia). Vision and hearing were impaired on clinical testing. Ocular movements were restricted in all directions in both the eyes and associated with decreased movements of facial muscles. There was generalized hypotonia and head unsteadiness. There was a soft tissue swelling in the right arm. Investigations showed Hb concentration 11 gm/dl, Total leukocyte count: 6400 cells/cmm, Differential count P60%, L 40%, ESR 11 mm/hr, Blood Urea Nitrogen 16 mg%, S. Creatinine: 0.8 mg%, Ca++ 8.4 mg%, S. Phosphorus: 4.8 mg% and S. Alkaline phosphatase: 239 IU. BAER and VEP were abnormal with no formation of waveforms bilaterally.

X-ray showed fracture of both humer with hyperplastic callus formation [Figure 1]. While in the hospital, the child developed fracture of left femur. X-ray skull and spine showed generalized osteoporosis with platybasia and basilar invagination. MRI brain showed non-formation of tentorium causing gross inferior descent of the occipital lobe up to the level of medulla. There was non-formation of bilateral cerebellar hemispheres and vermis with hypoplasia of the brain stem. There was diffuse paucity of myelination in bilateral cerebral hemispheres with poor differentiation of basal ganglia and thalamus. [Figures 2 and 3]. With this picture and a child presenting with multiple bone fractures with hyperplastic callus formation, generalized osteopenia and normal serum biochemical parameters, the diagnosis of osteogenesis imperfecta type V was made. The immediate differential diagnosis of a child with generalized osteopenia and multiple bone fractures include- rickets, renal tubular acidosis and hypoparathyroidism, all of which where excluded by normal serum biochemical parameters like calcium, phosphorus and alkaline phosphatase. The child was started on palindronate infusion 1 mg/kg/24 hr daily in order to inhibit bone resorption.
thus increasing bone mineralisation.

Osteogenesis imperfecta (OI) or Brittle bone disease is the most common genetic cause of osteoporosis.[1,6] The OI is caused by structural or quantitative defect in type I collagen. OI has the triad of fragile bone, blue sclerae and early deafness. OI type V is characterized by recurrent multiple fractures in a child following ambulation with hyperplastic callus formation at the sites of fracture.[11] This may be associated with calcification of introsseus membrane of forearm and a radidsense metaphyseal band. The diagnosis is usually clinical and may be confirmed by collagen biochemical studies using fibroblast cultured from a skin punch biopsy.

The recognized neurological complications of OI include cranio-vertebral junction anomalies like basilar invagination, syringohydromyelia, hydrocephalus and brainstem compression.[2] There is one case report of cerebellar hypoplasia associated with OI type 4.[1] This is the first case report of brainstem and cerebellar hypoplasia associated with OI type V. The proposed mechanism for cerebellar hypoplasia is intra-uterino vascular compromise due to associated cranio-vertebral junction anomalies.[3] In our patient all of the posterior circulation structures are hypoplastic supporting the hypothesis of vascular compromise.

Treatment with bisphosphonate drugs is effective in improving mobility and decreasing symptoms in many patients.[5,6] Intravenous pamidronate or oral alendronate improve quality of life and inhibit bone resorption thus increasing bone mineralisation. These agents decrease the frequency of fractures and amelionate bone pain.

References


Cicatricial ectropion due to herpes zoster ophthalmicus

Sir,

We present the clinical features and management of an unusual case of cicatricial ectropion of the upper lid secondary to herpes zoster ophthalmicus (HZO) in an immunodeficient patient.

Case History

An 83-year-old lady suffering from chronic lymphocytic leukaemia (CLL) was referred with a history of left herpes zoster ophthalmicus (HZO). The episode, which had been treated with systemic antivirals, had occurred 4 months prior to the referral. There was no intraocular involvement. On presentation to us, the vision was 6/24 on right and 1/60 on the left side. She had an uncomfortable red left eye. On examination [Figures 1 a, b, c, d], there was scarring on the left forehead extending down to the upper lid. The scar tissue had contracted resulting in upper lid eversion and retraction. She had poor lid closure and significant lagophthalmos. The exposed tarsal conjunctiva was keratinised and she had developed exposure keratopathy with a circumferential pannus, a hazy lustreless cornea and a central epithelial defect. Rest of the ocular examination was unremarkable.

In view of her age and general condition, she was offered a conservative tarsorrhaphy but she preferred reconstructive surgery. Under local anesthesia, a left upper lid wedge (18 by 12 mm) was resected to remove the cicatrix. The temporal upper lid was refashioned with a modified lateral tarsal strip. A full thickness skin graft from the left upper arm was excised, thinned and sized to reform the anterior lamella. Histopathology of the excised tissue revealed squamous metaplasia and a chronic active inflammatory infiltrate indicating post-inflammatory scarring.

Post-operatively the graft took well [Figure 2]. Full lid function was restored and the corneal epitheliopathy resolved.

Figure 3: X-rays showing fracture of right humerus with hyperplastic callus formation on right half and fracture of left femur with no callus on left half of figure 3

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