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CLEIDO-CRANIAL DYSOSTOSIS. CASE REPORT AND REVIEW OF SKELETAL ABNORMALITIES

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Cleido-cranial dysostosis is a rare hereditary disorder affecting intramembranous bone formation. It primarily affects the skull, clavicle and pelvis. Here, we describe a case presenting as a thoracic scoliosis. A review of the skeletal abnormalities is also discussed.
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Cleido-cranial dysostosis is a hereditary skeletal dysplasia characterised by deficient or imperfect ossification of intramembranous bone. The exact aetiology is unknown. A wide spectrum of skeletal abnormalities can be present. The disease is usually manifested at the age of two years. We report a case presenting with thoracic scoliosis.

THE CASE

An 8 year old Irish boy presented with a deformity of the back. On examination a rib hump was noticed with a curvature of the spine to the left side. Both shoulders were leveled and there was no evidence of decompensation. Neurological examination was normal. Both clavicles were not palpable.

X-ray of the spine showed left thoracic curve extending from T6 to L1 and measuring 300. There was no evidence of congenital vertebral anomalies. CT scan and MRI scan showed no evidence of cord tethering or syringomyelia. Chest x-rays revealed absence of both clavicles (Fig 1) and pelvic x-rays (Fig 2) showed delayed ossification of the bodies and pubic rami.

We elected to treat the child with thoracolumbar spinal orthosis (TLSO). Serial follow up x-rays showed further progression of his curve to 500 (Fig 3). Therefore, a subcutaneous Harrington rod instrumentation was performed. The future plan is to distract the spine every 6-12 months till maturity and then perform a formal posterior spinal fusion with a Harrington rod.

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DISCUSSION

Cleido-cranial dysostosis is a skeletal dysplasia characterised by an abnormal intramembranous ossification and affecting primarily the skull, clavicles and pelvis. Its classical description date back to 1897 when Marie and Sainton[1,2] named the condition hereditary cleido-cranial dysostosis. Both dominant and recessive inheritance has been described as well as occasional sporadic cases[3-5].

The typical patient is short, with large head and frontal bossing. The mandible is prominent with small maxilla. The anterior fontanelle is large and may never close completely. Depression of the sagittal suture has been described as the "Hot Cross Bun effect"[5]. Calvarial thickening may be present affecting the squamous portion of the temporal bone or the occipital bone[6]. Abnormal dentition is usually the reason why patients initially seek treatment. This might be

related to delayed eruption, non-eruption or incomplete development of permanent teeth[4].

Clavicular abnormalities give the patient the typical appearance of dropped shoulders with the ability of the patient to approximate them interiorly. These abnormalities vary from a small defect in one clavicle to total absence of both clavicles.

A cone-shaped thorax, present at birth and persists throughout life may be related to hypoplasia of delayed development of a segment of the vertebral body. Pelvic abnormalities include absence of ossification in the bodies and descending rami of the pubes, delayed ossification of the inferior rami of the ischia and coxa vara[7]. There have been scattered reports of various anomalies including congenital pseudoarthrosis of the tibia and femur[8,9] and pathological fractures[10].

Incomplete ossification of contours of embryonic vertebral arches account for various vertebral deformities like spina bifida, kyphosis, scoliosis, hemivertebra and cervical ribs. Patients with cleido-cranial dysostosis and progressive scoliosis should have CT scan and MRI scan to rule out the presence of syringomyelia[11].

The management of progressive scoliosis in cleido-cranial dysostosis presents a challenging problem. We believe that subcutaneous Harrington rod instrumentation is a good temporary option to control the rapid deterioration of the curve with the consideration of spinal fusion in the adolescence.

CONCLUSION

Cleido-cranial dysostosis is a rare skeletal dysplasia with a wide spectrum of skeletal manifestation. Scoliosis is a challenging problem to treat in these patients. It requires careful and thorough evaluation.

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