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When Dysmorphism Tells The Story: Incidental Diagnosis Of Cleidocranial Dysplasia In A 12 Year Boy.

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Abstract

Chromosomal deletions are an important cause of developmental delay and dysmorphic features in children. We report the case of a 12-year-old male child known case of beta thalassemia trait presented with speech delay and was incidentally found to have multiple dysmorphic features during pre-operative evaluation for adenotonsillectomy. Detailed clinical examination and multidisciplinary assessment were performed. Whole-genome sequencing revealed a deletion involving chromosome 6, consistent with cleidocranial dysplasia. Neuroimaging, Ophthalmology evaluation, Cardiac screening was unremarkable. This case emphasizes the importance of careful phenotypic assessment and genetic evaluation in children with developmental delay and dysmorphism.

Keywords: NIL

Introduction

Developmental delay associated with dysmorphic features often suggests an underlying genetic etiology. Chromosomal deletions constitute a significant proportion of such disorders and may present with variable and subtle clinical manifestations. Cleidocranial dysplasia (CCD) is a rare genetic condition characterized by craniofacial anomalies, dental abnormalities, and skeletal defects, most commonly linked to genetic alterations involving chromosome 6. Early recognition is essential to facilitate appropriate multidisciplinary care and genetic counselling. We report a case of chromosome 6 deletion presenting primarily with speech delay and dysmorphic features.

Case Summary

A 12-year-old male child, a known case of beta-thalassemia trait, was admitted for elective adenotonsillectomy. During preoperative evaluation, he was noted to have longstanding speech delay and multiple dysmorphic features. There was no history of perinatal insult, developmental regression, seizures, recurrent infections, or significant systemic illness. Family history was unremarkable, and there was no parental consanguinity.

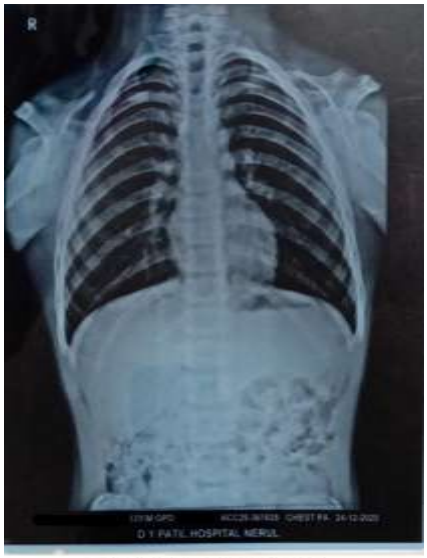
Physical examination revealed characteristic dysmorphic features including frontal and parietal bossing, hypertelorism, broad nasal bridge, low-set ears, prominent nasolabial folds, short philtrum, dental malocclusion, narrow drooping shoulders, sandal-gap deformity, and bilaterally shortened third and fourth toes. Growth parameters were appropriate for age, and systemic examination was otherwise normal.

Initial investigations, including MRI brain and ultrasonography of the kidneys, ureters, and bladder, were unremarkable. Given the presence of developmental delay and multiple dysmorphic features, a detailed genetic workup was undertaken. Whole-genome sequencing identified a chromosome 6 deletion, confirming the diagnosis of cleidocranial dysplasia.

Further skeletal evaluation demonstrated classical radiological features of cleidocranial dysplasia. Skull radiographs showed persistent widening of the sagittal

suture with multiple Wormian bones and delayed cranial ossification. Chest radiographs revealed bilateral clavicular aplasia and hypoplastic scapulae. Pelvic radiographs demonstrated widening of the pubic symphysis, hypoplastic iliac bones, and a characteristic "champagne-glass" pelvic inlet configuration. Cardiac and ophthalmological evaluations were normal.

The child was subsequently referred for multidisciplinary management, including genetic counselling, speech therapy, dental evaluation, and long-term orthopedic follow-up. This case highlights the importance of meticulous clinical examination and genetic evaluation in children presenting with developmental delay and subtle dysmorphic features, leading to the diagnosis of a rare skeletal dysplasia that might otherwise remain unrecognized.



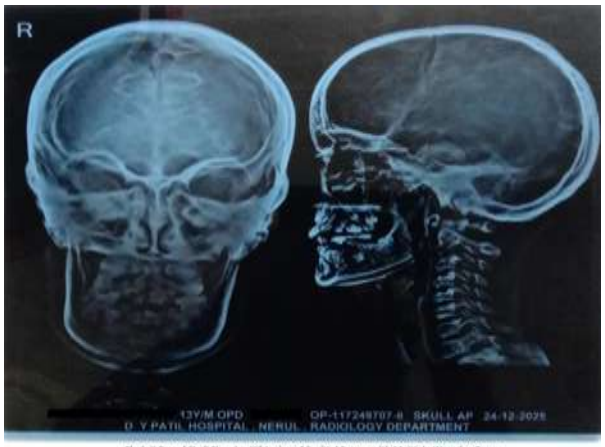


Fig 4: X Ray of Skull Showing Widening of Sagittal Suture with Multiple Wormian Bone

Fig 2: Image Showing Dysmorphic Features- Frontoparietal Bossing, Low set ears, Hypertelorism, Broad nasal bridge, Prominent nasolabial fold, Short Philtrum, Narrow drooping shoulder

Fig 3: Image Showing Sandal Gap

Discussion

This case highlights the importance of detailed clinical examination in children presenting with

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developmental delay, even when the presenting complaint is unrelated. Subtle dysmorphic features may be overlooked unless specifically sought. Chromosome 6 deletion are rare and can manifest with a wide phenotypic spectrum, including features overlapping with cleidocranial dysplasia. Genetic testing, particularly whole-genome sequencing, plays a crucial role in establishing the diagnosis, guiding further evaluation for associated anomalies, and enabling appropriate counselling of the family.

Early identification of such genetic conditions allows timely multidisciplinary interventions, including speech therapy, orthopaedic surveillance, dental care, and long-term developmental monitoring. This case underscores the need for a high index of suspicion and a systematic approach to evaluation in children with developmental delays.

Conclusion

Late diagnosis of cleidocranial dysplasia can occur when characteristic dysmorphic features are not recognized during early childhood. This case highlights the critical role of detailed phenotypic assessment in children with developmental delay and speech difficulties. Careful identification of dysmorphic features can provide vital diagnostic clues, facilitate timely genetic evaluation, and enable appropriate multidisciplinary management and counselling.

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