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8p Microdeletion Syndrome Presenting as Congenital Scoliosis and Facial Dysmorphism with Novel Anomalies

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Abstract | **Case Report**

8p deletion is a multisystem congenital disorder manifesting since early infancy with global developmental delay and other organ involvement. In our case the presentation was at 6 months with back deformity and developmental delay, dysmorphism and on further evaluation we found various structural anomalies. It needs a regular follow for early pick up of the associated comorbidities.

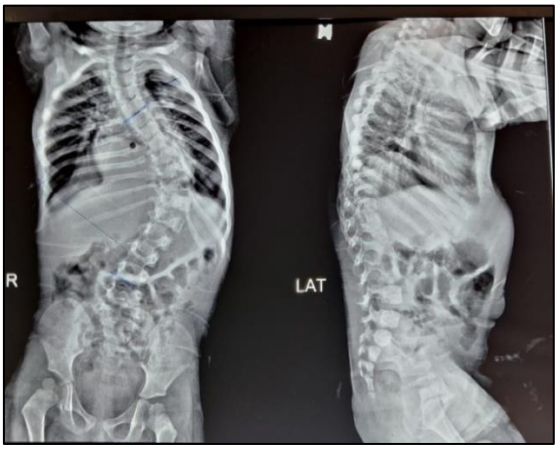
Keywords: 8p deletion, scoliosis, carpus callosum, butterfly vertebrae.

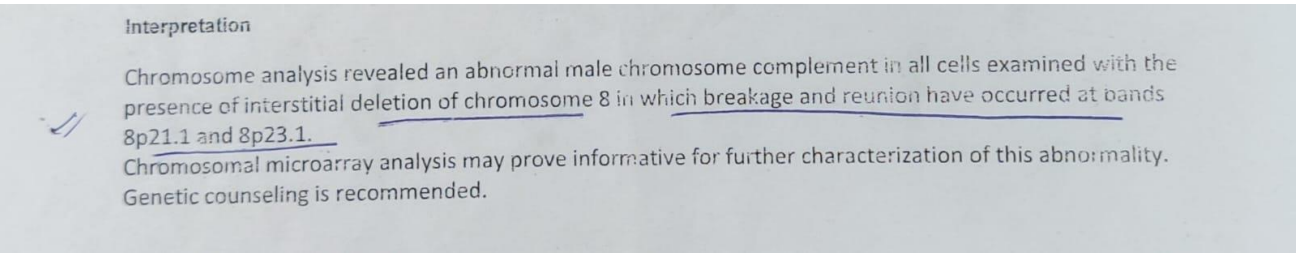
BACKGROUND

8p micro-deletion syndrome, is rare chromosomal anomaly clinically characterized by growth failure, mild to severe intellectual disability, severe developmental delay (psychomotor and speech development), facial dysmorphism, congenital malformations (CNS, skeletal, dental and CHD), hypotonia with tendency to later develop progressive hypertonia, and sensorineural hearing loss in few cases. Besides that these children may have immunological and endocrinal abnormality

CASE REPORT

A 1.5-year-old male child born out of 3rd degree consanguineous marriage, with a history of perinatal asphyxia and respiratory distress in the early neonatal period, presented to our OPD with deformity of back. The child failed to thrive with microcephaly, with global development delay (Language predominant). The child had facial dysmorphism- short, broad nose, wide depressed nasal bridge, retromicrognathia, cupid's bow of the upper lip, low set ears, upturned ear lobule, short neck, inverted nipple with normal limbs and genitalia, with no neurocutaneous markers. Spinal neuroimaging showed thoracic scoliosis with left convexity with butterfly vertebras (D5, D7, and D10). MRI brain showed the hypoplastic corpus callosum. Karyotyping showed interstitial deletion of Chr.8 in which breakage and reunion had occurred at bands 8p21.1 and 8p23. Karyotypes of parents were normal.





Interpretation

Chromosome analysis revealed an abnormal male chromosome complement in all cells examined with the presence of interstitial deletion of chromosome 8 in which breakage and reunion have occurred at bands 8p21.1 and 8p23.1.
Chromosomal microarray analysis may prove informative for further characterization of this abnormality.
Genetic counseling is recommended.

DISCUSSION

8p microdeletion should be considered a differential in a case presenting with congenital scoliosis with facial dysmorphism. Any child with 8p deletion should undergo detailed history and examination. Workup includes- Neuroimaging, 2d ECHO, USG Abdomen with KUB, chest radiography, spine radiography, dental, eye and hearing assessment. Regular follow-up should be done to assess growth and recognize any developing co-morbidity.

CONCLUSION

Parents and caregivers need to be adequately counselled about the disease process and complications. A vigilante watch on new symptoms or deterioration is need the hour. Correcting the deformities along with rehabilitating therapies improves the long term outcome.

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