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Cornelia De Lange Syndrome in Association with Diabetes Mellitus: A Case Report

Medical Science

KEYWORDS : Cornelia de lange syndrome,facial dysmorphism,growth and mental retardation,diabetes mellitus.

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ABSTRACT

Cornelia de lange syndrome(CdLS) is a rarely seen multisystemic developmental disorder syndrome characterized by facial dysmorphia(arched eyebrows,synophrys,depressed nasalbridge,anteverted nares,thin upper lip,long philtrum) along with musculoskeletal ,cardiovascular, gastrointestinal, genitourinary abnormalities with growth and mental retardation. We hereby present a case of this rare syndrome who along with other usual features presented to us with Type 1 diabetes mellitus. Association of Diabetes mellitus in CdLS has not been mentioned in any literatures or case reports till now.

Cornelia de lange syndrome

Cornelia de lange is a rare syndrome originally reported in 1933 by Cornelia de Lange. It is a rare syndrome and very few cases have been reported so far from India. The key diagnostic features are distinctive facial appearance, prenatal and postnatal growth deficiency, psychomotor delay, behavioral problems, and abnormalities of limb development.

Case Report

A 7year old male child of Indian origin,2nd by birth order, born of a non-consanguineous marriage presented to our hospital with chief complaints of polyuria from last seven days , not associated with any other complain and without any significant past and family history.Perinatal history was uneventful.He attained his gross motor milestones like peers and siblings but delayed fine motor, language and adaptive skills with poor school performance.

On physical examination,height was 111cm (less than -2SD of WHO growth charts) , weight was 16kg (less than -2SD), head circumference was 45cm (microcephaly),BMI was 12.9kg/m2 (less than 5th percentile). He had dysmorphic facial features like bushy eyebrows,synorphrys,long curly eyebrows,depressed nasal bridge,anteverted nares,thin upper lip,micrognathia,clinodactyly of fifth finger,syndactyly of 2nd and 3rd toe both foot,hypospadias. DQ in gross motor was >70%, around 50% in fine motor and around 30% in social and language domain. Rest of the systemic examination was unremarkable.

Lab analysis showed RBS of 435mg/dl, urine for sugar 4+, CBC,RFT,electrolytes and urine analysis were normal. ECHO showed trivial AR, radiographs of chest and forearm were normal.

Discussion

CdLS also called as brachman de lange syndrome is an autosomal dominant disorder, most cases are sporadic with marked variability in features.

Performance-These patients show marked growth retardation , moderate to severe mental retardation,hearing loss associated with speech delay,sometimes shows autistic behavior.

Facial features as microbrachycephaly, bushy eyebrows, synophrys, long curly eyelashes,depressed nasal bridge,anteverted nares,long philtrum, thin upper lip, high arched palate,late eruption of widely spaced teeth, micrognathia, hirsutism, micromelia, clinodactyly,simian crease, proximal implantation of thumbs,syndactyly of second and third toes,hypospadias,undescended testes.

Radiographic-dislocated/hypoplasticradial head,hypoplastic first metacarpal and fifth middle phalanx,shprt sternum with precocious fusion and 13 ribs Others-ocular abnormalities,short neck,GI problems. Occasional-seiures,congenital heart defects,diaphgramatic hernia,thrombocytopenia.

Conclusion

Coexistence of Type I DM with CdLS made it an interesting case to report due to importance of early recognition of the clinical condition and need of regular medical checkups and family counseling.

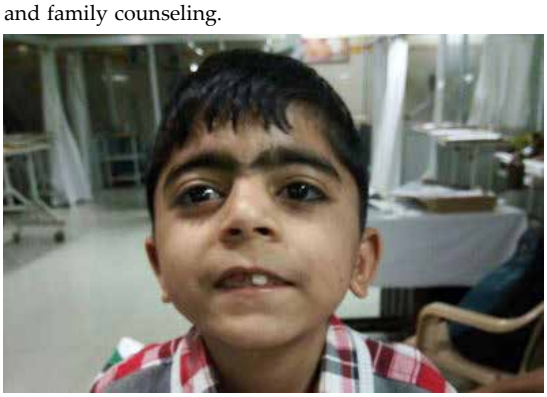

Fig.1: Facies in cornelia de lange syndrome showing Microrachy-cephaly, bushy eyebrows, synophrys ,long philtrum, thin upper lip and micrognathia.

Fig.2: Hands showing clinodactyly and proximal implantation of thumbs.

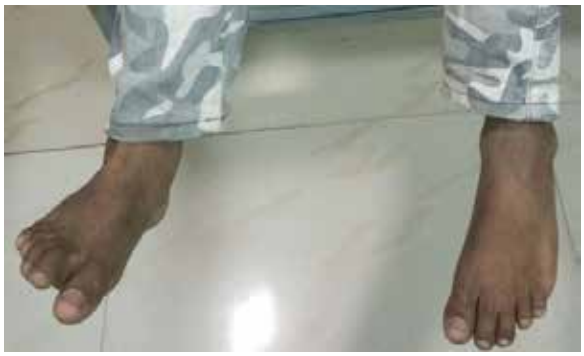

Fig.3:Feet showing syndactyly of second and third toes.

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