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Severe Intellectual Disability in a Case with Rubinstein-Taybi Syndrome: A Case Report

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Abstract: ***The chromosomal segment 16p13.3 microdeletion syndrome known as Rubinstein-Taybi syndrome (RSTS) is characterised by CREBBP gene mutations, a delay in the development of height and weight, distinctive facial features, broad and occasionally angulated thumbs and halluces, short stature, and mild to severe intellectual impairment. The present case report is of a female who came for disability certification and was incidentally identified with Rubinstein-Taybi syndrome.***

Keywords: Intellectual disability, Rubinstein - Taybi Syndrome, microcephaly, clinodactyly , genetic counselling

1. Introduction

The Rubinstein-Taybi syndrome is a rare congenital developmental disorder characterised by short stature typical in adulthood, moderate to severe learning difficulties, distinctive facial features (downslanted palpebral fissures, long lashes, high arched eyebrows, low-hanging columella, high palate, grimacing smile, talon cusps), broad and angulated thumbs and great toes usually involving the terminal phalanx[1]. In addition, the distal phalanx of thumbs and toes may also be misaligned on an abnormally shaped proximal phalanx(deltaphalanx). Clinodactyly may be present in 5th thumb[2]. Additional features of the disorder can include eye abnormalities, dental problems, obesity, heart and kidney defects. It is inherited in an autosomal dominant pattern. Most cases result from new mutations in the gene and occur in people with no family history. It occurs in 1 in 100000-125000 live births[2]. Very few cases are reported in literature from developing countries. Intellectual disability in these patients is almost constant but highly variable, with average IQ between 35-50.Language delay is present in 90% of the cases. Long term prognosis of these patients is generally good, but may vary due to range and severity of other health problems that may be present. Life expectancy generally does not seem to be affected.

2. Case Presentation

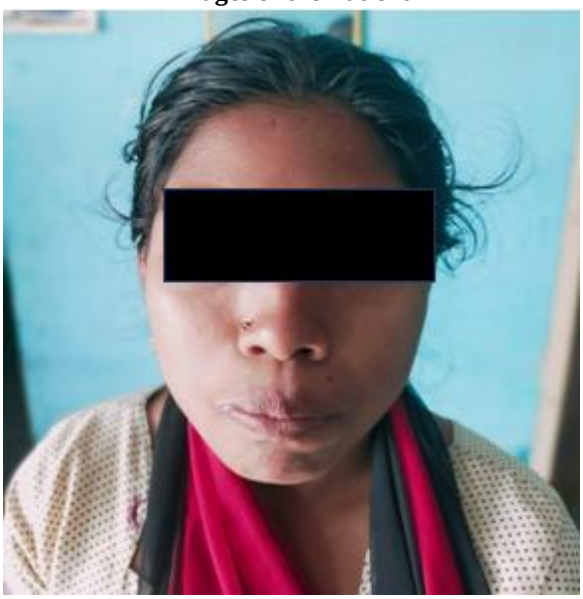
History:
A 20 years female, hailing from rural background, belonging to low socioeconomic status was brought to psychiatry OPD of SVRRGGH for disability certification. On evaluation, patient was born out of non-consanguineous marriage, FTNVD, low birth weight, with h/o speech delay and poor scholastic performance. Self care is maintained only with assistance. H/o seizures is present. No significant psychiatric history in the family. No h/o substance abuse.

On Examination:
Down-slanted palpebral fissures, long lashes, high palate, broad thumbs and great toes are present. Overcrowding of teeth is present.

Investigations:

- All routine blood investigations – CBC, LFT, RFT, S.Electrolytes, viral screening, thyroid profile, RBS were within normal limits.
- EEG showed abnormal report suggestive of right temporal epileptiform discharges with background slowing.
- MRI Brain showed normal study.

Images of the Patient



Images of the Patient

Diagnosis and treatment:
A provisional diagnosis of Rubinstein Taybi syndrome was made. Patient was started on anti-epileptic medications and was maintained on Tab. Carbamazepine 100 mg TID. Psycho- education was given to the family members.

3. Discussion

Rubinstein-Taybi syndrome (RSTS) is a rare syndrome, characterized by postnatal growth deficiency, microcephaly, characteristic facies, broad thumbs and big toes, and mental retardation.

Till date, over 1000 cases have been described in literature. RSTS is a prototype of diseases with genetic heterogeneity. Most cases of RSTS are sporadic, and families with more than 1 affected child are extremely rare.

The known genetic causes of RSTS are point mutations or microdeletions of CREB-binding protein gene (CREBBP) located on chromosome 16p 13.3 (in 50-60% affected patients), and of the EP300 gene encoding E1A binding protein p300 localized on 22q13.2 (in 5% patients)[3,4].The mutations causing RSTS are almost always de novo occurring in autosomal dominant fashion.

Adequate counselling of parents of a child with RSTS involves providing complete and comprehensive information on the syndrome itself, information regarding recurrence risks, and prenatal diagnosis[5].

4. Conclusion

The diagnosis of Rubinstein Taybi syndrome is primarily clinical and is based on characteristic phenotype that is often combined with a variety of somatic anomalies.

An early diagnosis facilitates appropriate genetic counselling and in planning the management.

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