Original Research Paper Pediatrics

A Case Report of A Rare Genetic Disorder

Dr C.Vimala Sarojini	Professor of Paediatrics in A.C.S. Medical College
Dr. R. Duraisami	Professor and HOD of Paediatrics in A.C.S. Medical College
Dr. Chandra K	Professor and HOD of General Medicine in A.C.S. Medical College
Dr. Rekha C	Assistant Professor of Paediatrics in A.C.S. Medical College
Dr Saranya L.R	Assistant Professor of Paediatrics in A.C.S. Medical College

KEYWORDS :

Background:
This is the report of a 15 month old child admitted for afebrile seizures. The clinical condition improved. The presence of post axial polydactyly in all four extremeties prompted us to get an opinion from an ophthalmologist and led to the diagnosis of 'BARDET BIEDL SYNDROME (BBS)'

. In BBS the onset of symptoms occurs in the first 10 years of life, poor night vision being the first symptom. This case is presented to highlight the importance of clinical examination. .

History:
15 month old female child was admitted in the paediatric ward on 13/06/2016 with a history of seizures

No H/O fever/ injury /previous episodes

3rd child born to 3◦ consanguineous parents delivered at term normal vaginal delivery.

Antenatal /natal/immediate postnatal periods uneventful

Head control at 5th month, sitting at 1 year, speaks double syllables

Child on breast feed till 6 month. Now child is taking family pot food.

On Examination
Head circumference - 45cm

Length- 70cm

Weight- 13kg

BMI – 26.5

AF admits tip of the finger

Squint +

Abnormal eye movements seen

No nystagmus

Polydactyly seen in all four extremities

Child able to sit without support

No focal neurological deficit

A diagnosis of seizure disorder with delayed mile stones with polydactyly was made

Ophthalmic examinations showed hypertelorism. Anterior chamber was normal.

Fundus: disc waxy pallor seen. Arterioles showed mild attenuation.

Bony spicule pigmentation in mid periphery similar to retinitis pigmentosa . Macular NAD .

This child with BMI of 26.5 (over weight)polydactyyl , mild development delay and retinal changes like retinitis pigmentosa was diagnosed as a case of BARDET BEIDL SYNDROME .

Investigations:
CBC, Calcium, LFT, RFT, Serum electrolytes all are Normal

USG-Abdomen – Normal

EEG – Normal

Genetic Study advised .Patient has not come for follow up with report

The careful clinical examination suggested the possibility of BARDET BEIDL SYNDROME. This case is reported to highlight the importance of clinical examinations.

DISCUSSION
The BARDET BEIDL SYNDROME is a multisystemic genetic disorder with autosomal recessive pattern of inheritance

Genetic mutations are noted in about 14 genes situated on the long arm of ch13 generally called as BBS genes which lead to the defective structure and function of celia.

1/4 of mutations are seen in BBS 1 gene

1/5 of mutations are seen in BBS 10 gene

In 25%of cases the causes is not known.

Incidence:
1 in 100000 to 1 in 160000 in NA & Europe.

Bedouins ,arab,new foundlands. 1:13500

Male: Female - 1.3:1

First found and described by BORDET&BIEDL in 1920.There is close similarity to the clinical features in syndrome described by Laurence moon biedl in 1865.

The syndrome is characterised by

Visual disturbance

Obesity

Polydactyl

Hypogonadism

Intellectual /learning disability

Renal abnormalities

Visual disturbance : 90-100% it is the major feature of BBS. This is due to dystrophy of cones and roads in retina leading on to defective night vision

Usually by 7 – 8 years (mid childhood)followed by blind spots in periphery. These blind spots merge to result in tunnel vision. By adousence loss of vision is ineritble .jerky eye movement and nystagmus are also seen in parents.

Obesity: obesity is also a characteristic feature of BBS .The truncal obesity begins in early childhood and is often associated with type II DM, hypertension and hypercholesterolemia

Polydactyly /syndactyly is a posvaxial and major presentation

Diagnosis during infancy is difficult. Not all characteristic features are present at birth except polydoctyly.

The visual impairment is noted at the age of 7 -8 years & and are legally blind by adolescence. Obesity appears after infancy which progresses and is even difficult to control.

Learning disabilities are noted only as the child grows.

Beales et al (1999) reported in a study of 109 patient .Average age of diagnosis was 9 years . Post axial polydactyly was seen at birth but obesity appeared around 8½yers.

Diagnosis

Usually made clinically. Presence of 3 major plus 2 minor criteria led to the diagnosis of BBS Clinically.

Genetic study is limited to research work.

Clinical criteria for diagnosis
Major criteria
1. Retinal degeneration 90 to 100 %

2. Obesity 72 to 92%

3. Polydatlyly at birth 63 to 81%

4. Hypogonadism 59 to 98%

5. Cognitive impairment 50 to 61%

6. Learning and speech difficulties and behavioural problems

7. Renal abnormalities 20 to50%

Minor criteria
1. Speech delay

2. Devolpmental delay

3. CHD

4. Barchy/syndactaly

5. Deafness

6. DM

7. Dental abnormality

8. Anosmia

4major or 3 major + 2 minor confirms the diagnosis of BBS clincally

Treatment
No definitive treatment

Diet / exercise / DM /HTN

Vision- use low vision aids/Orientation / mobility training

Education, special schools.

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