A CASE OF BARDET BIEDEL SYNDROME
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Abstract: Bardet-Biedl syndrome is a Ciliopathic human genetic disorder with variable expressivity and a wide range of clinical variability observed both within and between families. The main clinical features are Rod-cone dystrophy (atypical retinitis pigmentosa), Polydactyly, Obesity, Mental retardation, Hypogonadism in males, complex urogenital malformations in females, Renal anomalies and other secondary features. The syndrome is familial and is transmitted as an autosomal recessive trait. We like to report a case of Bardet Biedl syndrome because of its rarity in India as only less than 15 cases have been reported till now.

Keyword: Bardet Beidel syndrome, obesity, polydactyly, retinitis pigmentosa.

SELVI. SANGEETHA, 14 YEARS OLD GIRL WAS ADMITTED WITH C/O ABNORMAL WEIGHT GAIN AND VISUAL DISTURBANCES IN THE DIM LIGHT SINCE EARLY CHILDHOOD. H/O DECREASED SCHOOL PERFORMANCE + H/O EXTRA DIGITS OF BOTH HANDS AND FINGERS +

PAST H/O:
- PATIENT OPERATED AT THE AGE OF 4 MONTHS FOR UROGENITAL SINUS WITH ANTERIOR PERINEAL SINUS.
- GENITOSCOPY AT THAT TIME SHOWED VAGINAL ORIFICE AT BLADDER NECK REGION.
- POST SAGITTAL (PENA’S) AND RECTAL RANSPOSITION WITH VAGINO URETHROPLASTY WAS DONE.
- PATIENT WAS PUT ON DIET RESTRICTION BY HER MOTHER FOR PAST 1 YEAR FOR INCREASING WEIGHT GAIN.

BIRTH H/O:
- SHE WAS BORN OF 3rd DEGREE CONSANGUINOUS PARENTAGE. BIRTH WEIGHT – 3.5 KG DELIVERED BY CAESARIAN SECTION.

DEVELOPMENTAL DELAY PRESENT
- HEAD HOLDING – 1 YEAR OF AGE
- WALKING WITH SUPPORT – 1½ YEARS OF AGE
- WALKING WITHOUT SUPPORT – 2 YEARS OF AGE

FAMILY H/O:
- H/O SIMILAR SYMPTOMS PRESENT IN HER YOUNGER BROTHER
- ON EXAMINATION BMI: 25.80 kg/m2 CENTRAL OBESITY POLYDACTYLY PRESENT IN BOTH UPPER AND LOWER LIMBS
- BREAST NOT DEVELOPED ABSENT PUBIC AND AXILLARY HAIR
- HIGH ARCHED PALATE MILD MENTAL RETARDATION
- PERFORMANCE TESTING CANNOT BE DONE DUE TO VISUAL IMPAIRMENT
- EXTERNAL GENITALIA: LABIA MAJORA NORMAL LABIA MINORA – NOT WELL DEVELOPED
- RECTAL OPENING, VAGINAL ORIFICE AND ANAL OPENING – SEPARATELY MADE OUT
- VISUAL ACUITY 4/60 IN BOTH EYES
- FUNDS EXAMINATION: PIGMENTARY RETINOPATHY
- OTHER SYSTEM EXAMINATION WITHIN NORMAL LIMITS

POLYDACTYLY
POLYDACTYLY

INVESTIGATIONS:
CBC: HB% - 9.8 G% TC: 8800 CELLS/CMM DC: P65 L30 E5
RBC: 3.7 MILLION CELLS / CMM PLATELET: 1.1 LACK CELLS / CMM
RANDOM BLOOD SUGAR: 92 MGS / DL BLOOD UREA: 20 MGS / DL
SERUM SODIUM: 138 MEQ/L SERUM POTASSIUM: 3.5 MEQ/L
URINE ROUTINE EXAMINATION: NORMAL X-RAY CHEST - HEART AND LUNGS NORMAL ECG:
HEART RATE – 72/ MIN, SINUS RHYTHM, NO ST-T WAVE CHANGES
USG ABDOMEN:
LIVER – FATTY LIVER . 140 MM
SPLEEN: NORMAL . 110 MM
RIGHT KIDNEY: 105 * 48 MM . NORMAL ECHOTEXTURE . CMD +
LEFT KIDNEY: 102 * 41 MM . NORMAL ECHOTEXTURE . CMD +
BLADDER – NORMAL
UTERUS: 40 * 10 * 20 MM
OVARIES: RIGHT: 13 * 11 MM WITH FOLLICLE
LEFT: 21 * 14 MM WITH FOLLICLE
IMPRESSION: FATTY LIVER
THYROID PROFILE:
TOTAL T3 – 1.42 ( 0.8 – 2.0 ) NG / ML
TOTAL T4 – 10.9 (5.0 – 11.8 ) MIC/DL
TSH – 3.16 (0.3 – 5.5 ) MIC IU/mL
ECHO:
LVId: 3.7 CM LVId: 2.1 CM LV EF: 76.7 % NO RWMA OF LV
NORMAL LV SYSTOLIC FUNCTION IAS / IVS INTACT NORMAL
CHAMBERS / NORMAL VALVES NO PDA NO PHT
IMPRESSION: NORMAL STUDY
DISCUSSION:
The syndrome is familial and is transmitted as an autosomal recessive trait. Only 11 cases have been reported from India until 2009.
The detailed biochemical mechanism that leads to BBS is still unclear. At this moment, FOURTEEN genes that are responsible for the disease when mutated, have been cloned.
Genes involved include:
BBsome: BBS1, BBS2, BBS4, BBS5, BBS7, TTC8/BBS8, BBS9
Chaperone: BBS6, BBS10, BBS12
Other: ARL6/BBS3, TRIM32/BBS11

The gene products encoded by these BBS genes, called BBS proteins, are located in the basal body and cilia of the cell.

CLINICAL FEATURES TO DIAGNOSE ARE

PRIMARY FEATURES

- Retinitis pigmentosa
- Postaxial polydactyly
- Truncal obesity that manifests during infancy
- Learning difficulties
- Male hypogonadism and complex female genital urinary malformations
- Renal dysfunction, a major cause of morbidity and mortality.

SECONDARY FEATURES

- Speech disorder/delay
- Strabismus/cataracts/astigmatism
- Brachydactyly/syndactyly
- Developmental delay
- Polyuria/polydipsia
- Ataxia/poor coordination/imbalance
- Mild spasticity (especially lower limbs)
- Diabetes mellitus
- Dental crowding/ hypodontia/small roots/high arched palate
- Left ventricular hypertrophy/congenital heart disease
- Hepatic fibrosis

FOUR PRIMARY OR 3 PRIMARY AND TWO SECONDARY FEATURES ARE NEEDED TO DIAGNOSE BARDET BIEDEL SYNDROME.

Treatment is mainly focussed on symptoms:
Visual impairment – orientation and mobility training and use of a guide cane
Obesity is difficult to control but with a well balanced, specialized diet, a lot can be accomplished
Hypogonadism : hormone supplementation
Renal disorders – regular checkup

CONCLUSION:
CARDINAL FEATURES TO DIAGNOSE BARDET BIEDEL SYNDROME IN THIS PATIENT

PRIMARY:
OBESITY
MENTAL RETARDATION
GENITO-URINARY MALFORMATION
PIGMENTARY RETINOPATHY
POLYDACTYLY

SECONDARY:
DEVELOPMENTAL DELAY
HIGH ARCHED PALATE
DECREASED SCHOOL PERFORMANCE

References: