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Quiz: Images in Clinical Practice

Courtesy: Safder AMB

QUIZ – 1

Mr. X, aged 18 years, presented to endocrine OPD with the complaints short stature with arrested growth since 10 years of age, dimness of vision for 1 year, generalized body ache and mild bone pain for 1 month. He has no mental retardation. There were no features of hypogonadism. Abdominal examination revealed umbilical hernia & hepatomegaly.

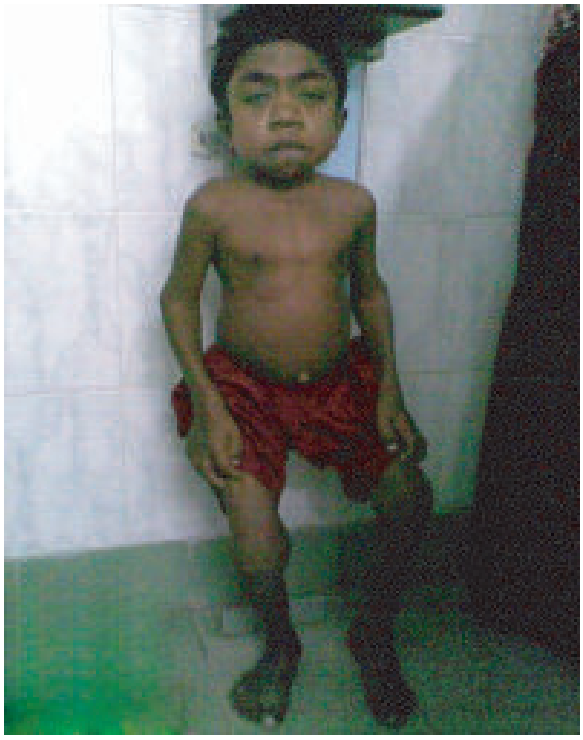

Fig.-1

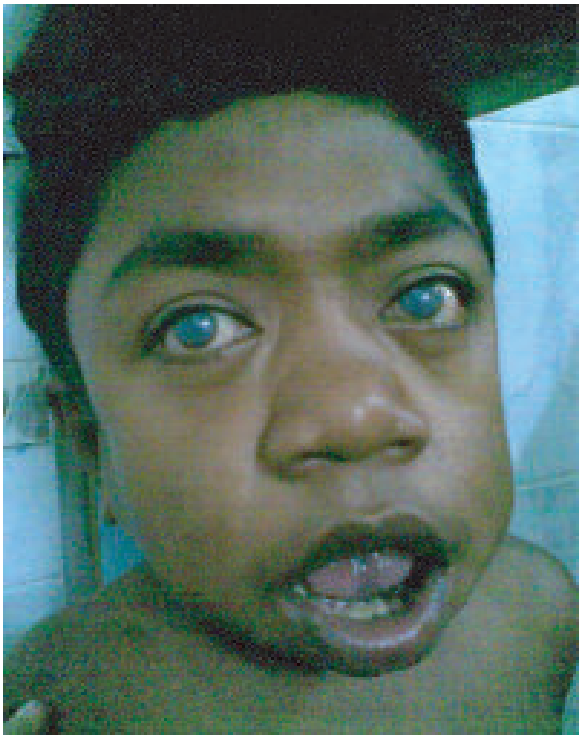

Fig.-2

What is the likely Diagnosis?

QUIZ – 2

A 27 year old male presented with progressive deterioration of vision since 8 years of age & Supernumerary digits of both hands and feet since birth. His achievement of developmental milestone was delayed and he was obese before age of 10 years .As he had learning difficulties; he quit school at early age. He had abnormal feeding behavior during his childhood with voracious appetite. His pubertal onset was delayed. He had cognitive impairment but his hearing and sense of smell were intact. What is the likely diagnosis?

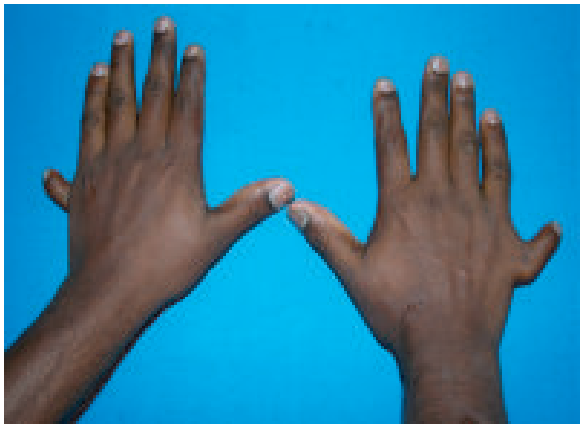

Fig.-1

Fig.-2

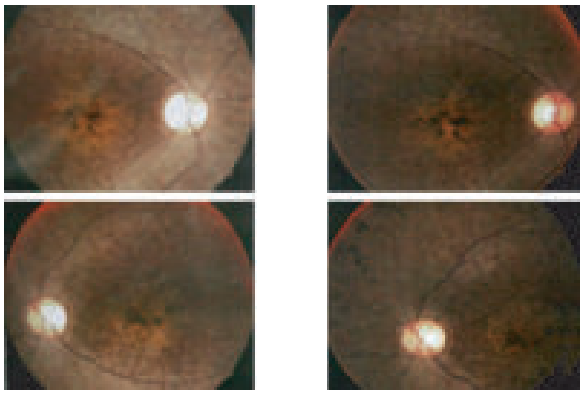

Fig.-3

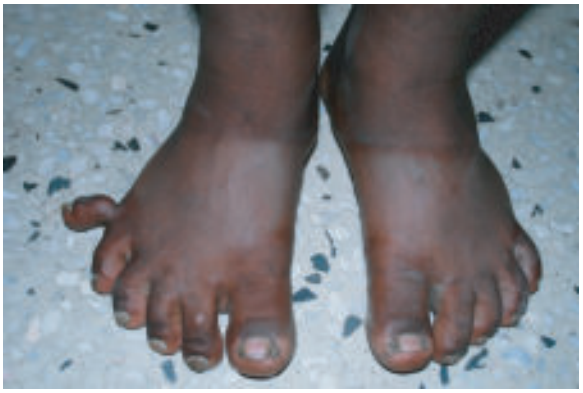

Fig.-4

Acknowledgement: Department of Endocrinology & Endocrine OPD, BIRDEM.

QUIZ – 1

Ans: Mucopolysaccharidosis-Type VI (Maroteaux-Lamy syndrome)

Picture: 1 Dysmorphic facial feature, short stature with flexion deformities in knee and elbow joints, umbilical hernia.

Picture: 2 Coarse facial features with flat nasal bridge, enlarged tongue, macrocephaly, prominent forehead, micrognathia, corneal opacity.

Review

Mucopolysaccharidoses are a group of metabolic disorders caused by the absence or malfunctioning of lysosomal enzymes needed to break down molecules called glycosaminoglycans (formerly called mucopolysaccharides). These are part of the lysosomal storage disease family. There are seven major subtypes. Most of them are autosomal recessive except MPS-I, which is an X-linked recessive disorder. Clinical features depend on the subtype & include the following:

- coarse or rough facial features (including a flat nasal bridge, thick lips, and enlarged mouth and tongue)
- short stature with disproportionately short trunk (dwarfism)
- dysplasia and other skeletal irregularities
- thickened skin
- enlarged organs such as liver or spleen
- hernia
- excessive body hair growth
- Short and often claw-like hands, progressive joint stiffness, and carpal tunnel syndrome can restrict hand mobility and function.
- Recurring respiratory infections are common, as are obstructive airway disease and obstructive sleep apnea.
- Many affected individuals also have heart disease, often involving enlarged or diseased heart valves.
- Neurological complications may include damage to neurons as well as pain and impaired motor function.
- Individuals may have normal intellect or may be profoundly retarded, may experience developmental delay.
- Many individuals have hearing loss, either conductive, neurosensitive or both.
- Communicating hydrocephalus occurs in some of the mucopolysaccharidoses.
- The eye's cornea often becomes cloudy from intracellular storage and glaucoma and degeneration of the retina also may affect the patient's vision.

Diagnosis often can be made through clinical examination and urine tests excess mucopolysaccharides are excreted in the urine. Enzyme assays are also used to provide definitive diagnosis of one of the mucopolysaccharidoses. Prenatal diagnosis can be done by amniocentesis and chorionic villus sampling. Currently there is no cure for these disorders. Medical care is directed at treating systemic conditions and improving the person's quality of life. Physical therapy and daily exercise may delay joint problems and improve the ability to move.

QUIZ – 2

Ans: Bardet Biedl Syndrome or Laurence Moon Bardet Biedl Syndrome (LMBBS)

Picture: 1, 4 polydactyly.

Picture: 2 Average body built.

Picture: 3 Retinitis pigmentosa.

Review

Bardet BiedI Syndrome is characterized principally by red- cone dystrophy, with childhood onset visual loss preceded by night blindness ; postaxial polydactyly, truncal obesity that manifests during infancy and remains problematic throughout adult; specific learning difficulties; male hypogenitalism and complex female genitourinary malformations ; and renal dysfunction. In 1865 Laurence Moon reported the first case of obese, visually impaired girl with intellectual disabilities. In 1920s , two independent reports by George Bardet and Artur Biedl describing the additional characteristics of polydactyly and hypogenitalism , all these remain the cardinal feature of BBS, but further manifestations of the disease have since been recognized.

Primary symptoms (most common in the syndrome)

- Visual impairment caused by rod-cone dystrophy, often diagnosed as retinitis pigmentosa
- Extra fingers and/or toes (polydactyly)
- Truncal obesity
- Learning disabilities
- Underdeveloped genitals in males (hypogonadism)
- Kidney abnormalities

Secondary symptoms (may also occur)

- Delayed development
- Speech problems
- Poor coordination, clumsiness
- Type 2 diabetes
- Difficulty controlling muscles (ataxia)

For diagnosis, four primary symptoms should be present, or three primary plus two secondary symptoms. There is no specific test to detect the presence of LMBBS. Treatment is focused on the symptoms.