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The Medical Bulletin

In Syndrome

Bardet - Biedl syndrome (BBS) / Laurence-Moon-Biedl-Bardet

Definition :

It is a multisystem non-motile ciliopathy.

Epidemiology : - 1 in 100,000

Causes :

- It is typically inherited in an autosomal recessive pattern.
- Mutations in BBS1 to BBS18 gene accounts for about 70%–80%.
- Mutations in BBS genes lead to problems with the structure and function of cilia.
- Defects in these cell structures probably disrupt important chemical signaling pathways during development and lead to abnormalities of sensory perception.

Clinical Features :

- A clinical diagnosis of BBS is made by the presence of either four major features or three major features and two minor features.

Major features :

- Retinal cone – rod dystrophy, Central obesity, Postaxial polydactyly, Cognitive impairment, Hypogonadism & genitourinary abnormalities, Kidney disease.
- Minor features : Neurologic abnormalities - (DD, epilepsy, behavior/psychiatric abnormalities) , Olfactory dysfunction, Oral/dental abnormalities, Cardiovascular & other thoraco-abdominal abnormalities, Gastrointestinal abnormalities (hirschsprung disease, inflammatory bowel disease, celiac disease, liver disease), Endocrine/metabolic abnormalities(metabolic syndrome, subclinical hypothyroidism, T2DM, polycystic ovary syndrome).

Diagnosis :

- Generally a clinical diagnosis.
- Genetic testing may help confirm the diagnosis for some patients (e.g., individuals with certain BBS1 and BBS10 gene mutations)
- As diagnosis is based on clinical findings and BBS is associated with variable expression of the classical features, some patients may not have a clear diagnosis for many years.

Treatment :

- The management of patients is multidisciplinary.
- Treatment is basically symptomatic and complete recovery is not possible.

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