Translation from Romanian

Bardet-Biedl Syndrome

DEFINITION:

→ **Bardet-Biedl syndrome (BBS)** is a ciliopathy (a genetic disease that affects the cilia of cells, their anchoring structures, basal bodies, or ciliary function) with multisystemic involvement.

PREVALENCE: in Europe it is estimated at **1/125.000 – 1/175.000**

CLINICAL ASPECTS:

→ The disease is characterized by a combination of **clinical signs:** obesity, retinitis pigmentosa, postaxial polydactyly, deafness, polycystic kidney disease, hypogenitalism and learning disabilities, many of which occur a few years after the onset of the disease.

→ The clinical expression is variable, but most patients show most clinical signs during the disease.

→ **Retinitis pigmentosa** is the only constant clinical sign after childhood.

→ It can be associated with several other manifestations: diabetes, hypertension, congenital heart disease and Hirschsprung's disease

GENETICAL ASPECTS:

→ The observed broad clinical spectrum is associated with significant genetic heterogeneity. To date, the mutations in **14 different genes (BBS1 to BBS14)** have been identified as responsible for this phenotype. These genes encode the proteins involved in the development and functioning of primary cilia. The absence or dysfunction of BBS proteins causes ciliary abnormalities in organs, such as the **kidney or eye.**

→ However, the relationship between symptoms and ciliary dysfunction remains unknown for some of the clinical manifestations of BBS.

→ The disorder is transmitted mainly in an **autosomal recessive** manner, but in some cases **oligogenic transmission** has been reported.

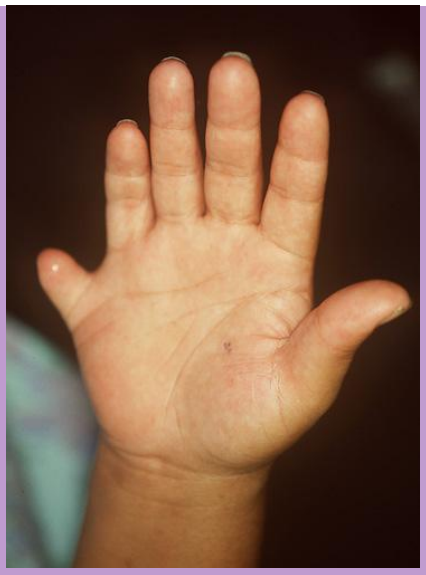
THE DIAGNOSIS can be confirmed by molecular analysis, allowing adequate genetic counseling for family members and possible prenatal diagnosis.

THE TREATMENT –requires a multidisciplinary approach

PROGNOSIS: Renal abnormalities are the main life-threatening conditions because they can lead to end-stage renal failure and may require kidney transplantation.

→ The progressive loss of vision due to **retinal dystrophy,** along with moderate intellectual deficit (when present), behavioral abnormalities, hypomimia and obesity can affect the social life of these patients.

Translation from Romanian



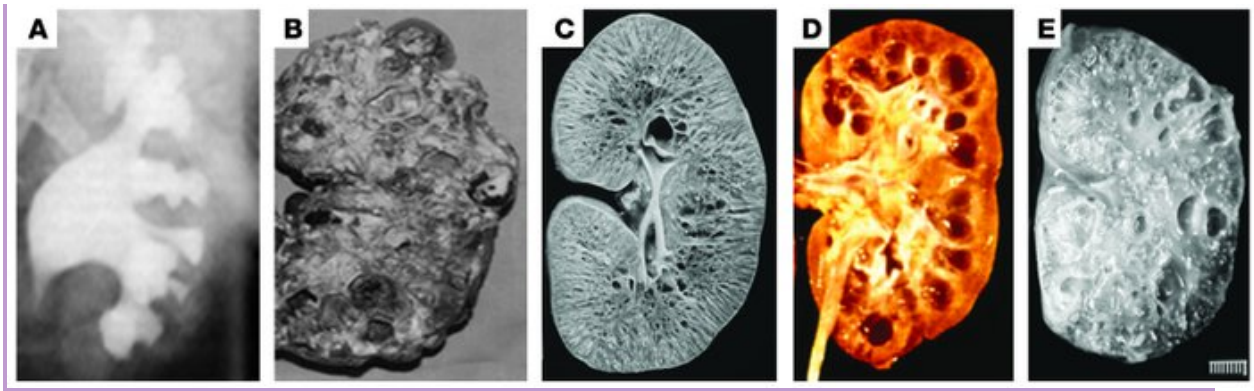
Img. 1: Post-axial polydactyly in Bardet-Biedl syndrome

Source: https://emedicine.medscape.com/

Img. 2: Facies in Biedl-Bardet syndrome

Source : https://www.fdna.com/blog/tyler-laurens-journeys-bardet-biedl-syndrome/

Translation from Romanian



Img. 3: Polycystic kidney in Bardet-Biedl syndrome

Source : https://www.jci.org/articles/view/37041/figure/3

Text source: https://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN

I, BURA TEODORA - NATALIA, certified interpreter and translator for English, by virtue of the authorization no. 35530/20.03.2013, issued by the Ministry of Justice of Romania, hereby certify the accuracy of the translation from Romanian into English.

Traducător și Interpret Autorizat
Bura Teodora-Natalia
ENGLEZĂ
Aut. M.J. 35530/2013