

# Bardet-Biedl Syndrome

## Main Features

- Dysfunction of Brain, eye, kidneys and genitalia

## Eye Findings

- Retinal degeneration (almost 100%)
  - Rod-cone dystrophy with early macular involvement
    - Pigmentary changes in macula first
    - ERG
  - Symptoms in order of presentation
    - Nyctalopia age 4-8 years
    - Progressive peripheral vision loss
    - Decline of color vision
    - Legal blindness by 20-30 years old
- Strabismus
- Cataracts
- Astigmatism

## Bardet-Biedl Syndrome Retinopathy Clinical Grading System

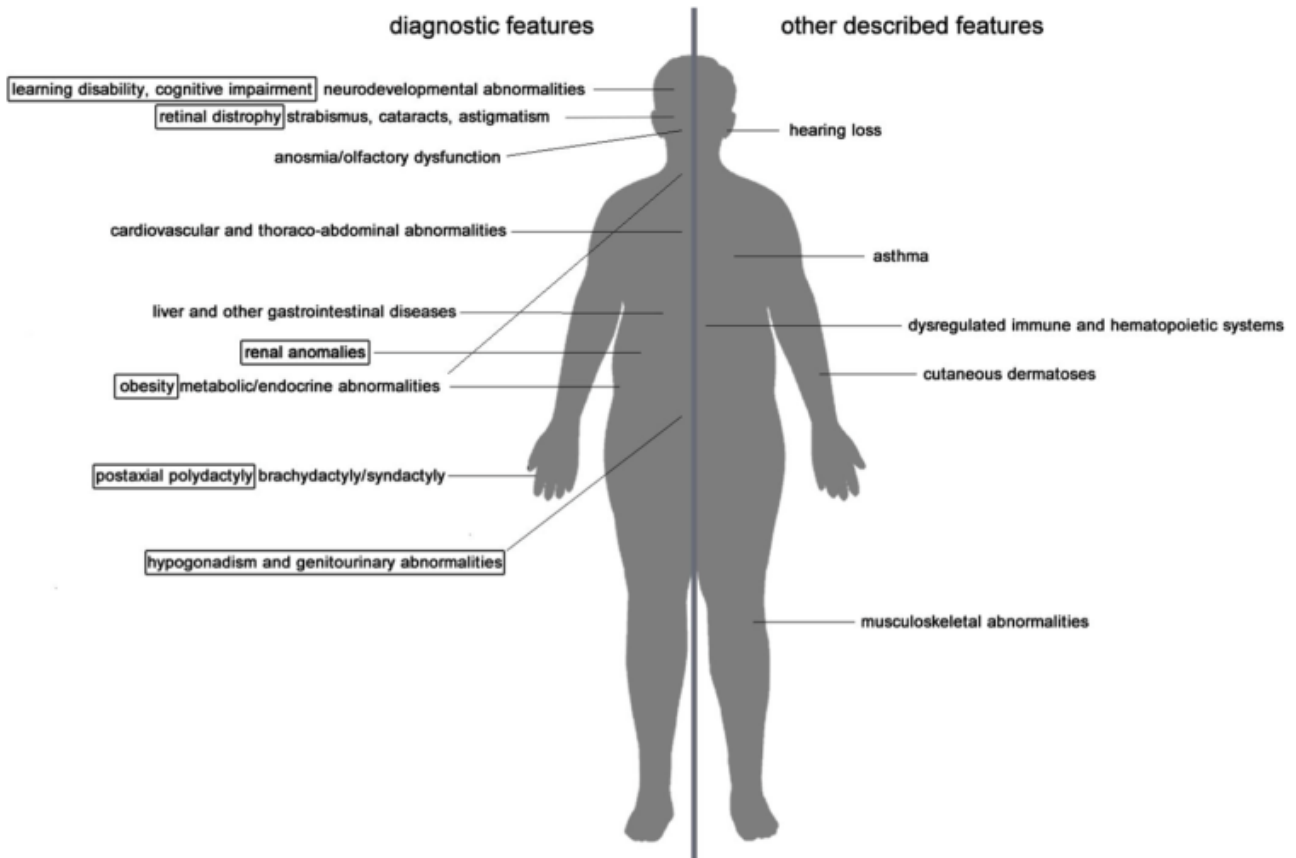
Grade	Description	Subtypes	Clinical Features
0	Normal		None
1	Preclinical Retinopathy		No fundoscopic evidence of retinopathy but evidence of reduced visual function (acuity, visual fields, ERG)
2	Clinical Retinopathy	2c	Central disease with foveal atrophy
		2p	Peripheral disease with pigmentary reinopathy
		2g	Global disease with central and peripheral involvement but no features of stage 3
3	End-stage retinopathy		Advanced central and peripheral involvement, which may include extensive scarring, vascular attenuation, choroidal atrophy, and significant disc pallor

Adapted from Dennison AK et al. *Retina* 2014;34:2282-2289

## Other Findings

- Obesity

- Postaxial polydactyly
- Renal anomalies
- Learning disability
- Hypogonadism and Genitourinary abnormalities
- Cardiac defects



**Figure 2** Major clinical features of BBS patients. On the left side diagnostic clinical features are listed (primary features outlined with rectangles). On the right side, less described clinical traits.

From resource 1 below

## Etiology

- Non-motile ciliopathy- dysfunction of the primary cilium.
- Autosomal Recessive

## Resources

1. [Bardet-Biedl Syndrome: Current Perspectives and Clinical Outlook. Melluso A et al. Therapeutics and Clinical Risk Management. 2023;19:115-132](#)
2. [Evaluation of Visual Function and Needs in Adult Patients with Bardet-Biedl Syndrome. Denniston AK et al. Retina 2014;34:2282-2289](#)

syndrome

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