Genetics Uncoded:

Facts about

Bardet-Biedl Syndrome (*BBS1*-related)

What Your Test Results Mean

Carriers typically show no symptoms of BBS1-related Bardet-Biedl syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Bardet-Biedl Syndrome Explained

Bardet-Biedl syndrome is an inherited condition of progressive vision loss, obesity, extra digits, intellectual disability, kidney disease, and abnormalities of the genitalia. There is no cure for Bardet-Biedl syndrome, but symptomatic management may alleviate some health issues. Individuals with Bardet-Biedl syndrome can live well into adulthood, but kidney disease is a major cause of early death.

How the Genetics Work

Bardet-Biedl syndrome is an autosomal recessive disorder caused by variants in a variety of genes. Variations in the *BBS1* gene cause of over 20% of Bardet-Biedl syndrome cases. In general, individuals have two copies of the *BBS1* gene. Carriers of Bardet-Biedl syndrome have a single variant in one copy of the *BBS1* gene, while individuals with Bardet-Biedl syndrome have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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