

17 January 2023

Bardet-Biedl Syndrome (BBS)

Could i please request the following information?

If you treat Bardet-biedl Syndrome, how many unique patients have you treated in the last 12 months?

What % of these patients get a genetic diagnosis prior to them being referred onto specialist centres?

If you do not treat Bardet-biedl Syndrome, could I request where these patients are referred to?

Belfast Health and Social Care Trust has reviewed this Freedom of Information request from a whole-Trust perspective. Bardet-Biedl Syndrome (BBS) encompasses a range of ciliopathy disorders caused by mutations in a large number of genes. More information is available at [ORPHA.net](https://orpha.net). (Bardet-Biedl syndrome: ORPHA:110)

The Northern Ireland Regional Genetics Service is based at Belfast City Hospital and provides a regional service covering all of Northern Ireland. Patients with BBS typically have a variety of clinical needs and, consequently, attend multiple specialties, some of which are based in Belfast Trust and others delivered in their local Trust of residence. Others attend the National BBS Clinics at Birmingham Women's Hospital and Guy's Hospital in London on an ad hoc basis, usually after referral from Clinical Genetics. As far as we can tell, all have had molecular testing carried out to confirm the precise genetic basis of their condition – this is generally conducted through the Northern Ireland Regional Genetics Laboratories on a send-away basis.

We have 24 patients coded on our IT system as having BBS. Many of these patients see a wide range of professionals within Belfast Trust.

In answering the questions below, the service have counted any logged Trust contact in the calendar year 2022 as being within the "last 12 months".

- **If you treat Bardet-biedl Syndrome, how many unique patients have you treated in the last 12 months?**

There were 19 patients treated/had clinical contact in the calendar year 2022.

- **What % of these patients get a genetic diagnosis prior to them being referred onto specialist centres?**

100%. Genetic testing has evolved over the years; some older patients would have been referred on the basis of a clinical diagnosis, with subsequent molecular testing. Currently, all patients being referred to the National Clinics will have a molecular diagnosis established prior to referral.

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- **If you do not treat Bardet-biedl Syndrome, could I request where these patients are referred to?**

Not applicable. Although some patients have either a one-off or follow-up appointment at the National BBS Clinics, this is generally due to patient preference.

All clinical services are available in Northern Ireland and the clinical team at the Northern Ireland Regional Genetics Service are expert in management of a wide range of complex genetic disorders, including BBS.