

FOI NUH 62182

Q1	If you treat Bardet-biedle Syndrome, how many unique patients have you treated in the last 12 months?
A1	<p>I can confirm that the Trust holds information that falls within the description specified in your request. However, the information is not held in an easily retrievable format. The code for Bardet-Biedl Syndrome is Q87.8.</p> <p>As this is a .8 this would include certain other congenital malformation syndromes including Alport and Zellweger and would require the retrieval and manual audit/review of individual records in order to extract the information requested.</p> <p>We estimate that the cost of complying with your request would exceed the appropriate limit of £450. The appropriate limit is set by the Secretary of State in SI 2004 No 3244 - The Freedom of Information and Data Protection (Appropriate Limit and Fees) Regulations 2004 has a limit of £450 for Public Authorities. This represents the estimated cost of one person spending over 18 hours (at rate of £25 per hour) in determining whether the Trust holds the information, and in locating, retrieving and extracting information</p>
Q2	What % of these patients get a genetic diagnosis prior to them being referred onto specialist centres?
A2	See A1
Q3	If you do not treat Bardet-biedle Syndrome, could I request where these patients are referred to?
A3	<p>We do manage BBS patients from the ophthalmology point of view and probably see one or two patients per year who usually have a diagnosis already confirmed. However, since these patients need a multidisciplinary approach, they are often referred to National Services for rare disease in Birmingham, GOSH and Guy's and Thomas where they see multiple specialists in one day. We understand the majority of the referrals to those centres come from GPs, geneticists or paediatricians.</p>