Royal Devon University Healthcare NHS Foundation Trust

Title:

Chromosome Abnormalities / 4q Deletion Syndrome

Reference Number: RDF1556-23
Date of Response: 26/06/2023

Further to your Freedom of Information Act request, please find the Trust's response(s) below:

Questions:

1) Of all patients found to have one or more genetic abnormalities, what percent have been found to have the following symptoms:
a) Feeding issues in infancy.
b) Failure to Thrive.

The Trust has considered your request. It is not clear from the question whether information is requested just for patients with Chromosome 4q deletion or all patients with a genetic disorder; **however, regardless of that** it is not possible to provide data to the requested level of detail as a full paper notes review would need to be carried out to confirm the detail of the genetic abnormality and then ascertain if any of the symptoms listed were noted. This would take considerable time (over the allocated 18 hours for any FOI response) and would need to be carried out by a Consultant Clinical Geneticist, at a cost far exceeding the appropriate limit.

12. — (1) Section 1(1) does not oblige a public authority to comply with a request for information if the authority estimates that the cost of complying with the request would exceed the appropriate limit.
The appropriate limit of £450 represents the estimated cost of one person spending two and a half days This would entail Trust staff: -

- Determining whether the information is held.
- Locating the information, or a document containing it.
- Retrieving the information, or a document containing it.
- Extracting the information from a document containing it.

To carry out the above would exceed the appropriate cost limit as set out in Section 12 (1) of the Freedom of Information Act 2000 and is therefore exempt.

2) 4q Deletion Syndrome, or simply 4q Syndrome is defined in medical literature and studies as "a rare chromosomal disorder where a portion of the 4th chromosome long arm (4q) is deleted."

Please disclose how many patients have met these criteria from all genetic testing done over the past 3 years.

The Trust has considered your request. It is not possible to provide data to the requested level of detail from the clinical genetics system, Trak Gene. The system cannot be searched for 4q deletion, only for chromosome 4 abnormality, and whilst some of those may be 4q, some will be other deletions or duplications. There will also

be some 4q deletions coded under chromosome deletion or just chromosome abnormality. 35 pts with chromosome 4 abnormality can be identified from Trak Gene; however, a full paper notes review would need to be carried out to confirm the detail underpinning the diagnosis and when the diagnosis was made. As above, this would take considerable time and would need to be carried out by a Consultant Clinical Geneticist, at a cost far exceeding the appropriate limit. It would also result in an incomplete dataset due to implementation of Epic from Oct 2020, as we do not have the facility to search Epic for these criteria.

In a bid to assist under Section 16 of the Freedom of Information Act, the provision of genomic testing within England is based off a National Genomic Test Directory (https://www.england.nhs.uk/publication/national-genomic-test-directories/), covered by 7 "genomic laboratory hubs" covering distinct geographical regions. This testing would be offered within the South West region by North Bristol Trust under this arrangement.

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3)
Please disclose all information you hold on 4q Deletion Syndrome, sometimes known as 4q Syndrome. This is defined in medical literature and published studies as "a rare chromosomal disorder where a portion of the 4th chromosome long arm (4q) is deleted."

The Clinical Genetics service use information from DECIPHER, Unique patient information leaflets https://rarechromo.org/disorder-guides/ , and if seeing a patient would carry out a literature search using PubMed and review relevant papers. Papers are read online; they are not printed or retained so cannot be shared with the requester.