

Title: Chromosome Abnormalities

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Reference Number: RDF1078-22

Date of Response: 22/12/22

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

*Please provide the information below for microdeletions and microduplications in the chromosomal bands/locations listed below.*

*Chromosome Bands*

*2p22.1*

*4q27 - 4q28.1*

*4q28.1*

*4q28.2*

*4q28.3*

*7q11.21*

*15q11.1 - 15q11.2*

*Information Requested:*

- 1) *Symptoms documented to be found in patients with chromosomal or genetic abnormalities in these regions.*

Answer: The Trust has considered your request; however, the information is not held in a manner to provide you with the information requested, this would require the manual extraction and manipulation of information from individual records. To carry out this work would exceed the appropriate cost limit as set out in Section 12 (1) of the Freedom of Information Act 2000 and is therefore exempt.

Under the Freedom of Information Act 2000 Section 12 (1) and defined in the Freedom of Information and Data Protection (Appropriate Limit and Fees) Regulations 2004, a public authority is not obliged to comply with a request for information if it estimates that the cost of complying would exceed the appropriate limit. The limit of £450 represents the estimated cost of one person spending two and a half days in determining whether the Trust holds the information, locating, retrieving and extracting that information.

- 2) *Any medical conditions or syndromes which occur as a result of abnormalities in these regions.*

Answer: Please see response to question 1.

- 3) *Whether the Trust uses and/or contributes to the DECIPHER genomic database.*

Answer: Yes.

- 4) *Details of what, if any, genomic database(s) the Trust use to identify chromosomal/genetic abnormalities and conditions identified in patients.*

Answer: DECIPHER.

5) *Details of how the Trust reports, documents and shares information about chromosomal/genetic abnormalities detected in patients. This applies to internally within the Trust and NHS, and also with the wider genomics community across the world.*

Answer: Results are shared with individual patients and uploaded to DECIPHER.