

DDX3X Gene

Reference Number: RDF1861-23

Date of Response: 06/10/23

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

Please be aware that the Royal Devon University Healthcare NHS Foundation Trust (Royal Devon) has existed since 1st April 2022 following the integration of the Northern Devon Healthcare NHS Trust (known as Northern Services) and the Royal Devon and Exeter NHS Foundation Trust (known as Eastern Services).

I am writing to you under the Freedom of Information Act 2000 to request the following aggregated information from your genetics department.

Number of individuals diagnosed with a mutation in the DDX3X gene:

- 1. Total population diagnosed with a DDX3X mutation, identified as DDX3X and/or related terms (DDX3X, DEAD-box helicase 3 X-linked, DBX, DDX3, HLP2, DDX14, CAP-Rf, MRX102, MRXSSB, DEAD (Asp-Glu-Ala-Asp) box helicase 3, X-linked, DEAD (Asp-Glu-Ala-Asp) box polypeptide 3, X-linked, DEAD/H (Asp-Glu-Ala-Asp/His) box polypeptide 3. OMIM code 300160, Orphanet 358713, NCBI Gene ID 1654, HGNC:2745.).*

Answer: We have identified reportable pathogenic variants in ≤5 patients within the date range 2018 to 2022.

- 2. Number of individuals diagnosed in each year*

Answer: Please see below.

- 2018 – ≤5
- 2019 – ≤5
- 2020 - Nil
- 2021 – ≤5
- 2022 – ≤5

In accordance with Section 40 (2) of the Freedom of Information Act 2000, we are unable to provide figures where the number of patients is less than or equal to five and could risk the identification of those patients and breach Caldicott principles. In these cases ≤5 is used to indicate that a figure between 1 and 5 is being suppressed.

This follows NHS Digital (formerly HSCIC) analysis guidance (2014) which states that small numbers within local authorities, wards, postcode districts, CCG's providers and Trusts may allow identification of patients and should not be published.

3. *Year of Birth of individuals diagnosed with DDX3X*

Answer: The Trust has carefully considered your request. However, it declines to provide the specific details of the information requested under Section 40 (3a) of the Freedom of Information Act.

The Trust recognises that whilst the information requested does not itself identify the patient(s), the question asks for very specific and focused details, and due to the low number of cases involved to provide this detail would create a potential for the patient(s) concerned to be identified and it is likely the Trust would be in breach of the Data Protection Act principles.

The Trust believes that the release of such sensitive information meets the definition of personal data and disclosing the information would contravene one of the data protection principles set out in Article 5 of the UK GDPR. As such release of the information would be likely to cause distress to the individuals concerned.

4. *Age of individuals when diagnosed.*

Answer: Please see response to question 3.