

Timothy Syndrome - CACNA1C Gene

Reference Number: RDF1603-23

Date of Response: 27/06/23

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 seeking information under the FOI Act regarding Timothy Syndrome and individuals with a change to a gene called CACNA1C.

My question is:

- 1. How many individuals were identified to have a CACNA1C gene change as a result of genetic testing during the year 2022 and during the year 2021?*

When a genetic change is found it is categorised according to the ACGS Guidelines (Association for Clinical Genomic Science). Ideally your response to how many individuals identified would indicate the classification of the findings with regard to number of individuals ie. Pathogenic, Likely pathogenic, Variant of Uncertain Significance, Likely benign and Benign.

In your answer, it would be helpful to know within the National Genomic Test Directory which panels were used to identify these individuals and your catchment area please. To assist, a current list of panels for CACNA1C can be found here:

<https://panelapp.genomicsengland.co.uk/panels/entities/CACNA1C>

Answer: 6 variants across ≤ 5 individuals (2 pathogenic, 4 likely pathogenic). Variants were identified via gene-agnostic approach (i.e. gene panel independent). Four of these were found in 2021 and two in 2022.

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authorities, wards, postcode districts, CCG's providers and Trusts may allow identification of patients and should not be published.