

Ref. No: 764
Date: 26-May-2023
Subject: Timothy Syndrome Alliance

REQUEST

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>

RESPONSE

The Clinical Director of Paediatrics has confirmed that we do not have this service at this Trust.