

Our Ref: 229/2023
MAY 2023

Re: Your request made under the Freedom of Information Act 2000

My question is:

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?

0 All our genetic test for children are performed at Sheffield Children's Hospital.

<https://www.sheffieldchildrens.nhs.uk/contact-us/>

If you are unable to find the information you require on our website you can make a request under the freedom of information act by the following methods:

1. Email your request to scn-tr.schfoi@nhs.net
2. Write to Carolyn Sampson, Senior Information Governance Officer, Sheffield Children's NHS Foundation Trust, Western Bank, Sheffield S10 2TH.

All Freedom of Information (FOI) requests will be responded to within 20 working days.

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>