Freedom of Information Act 2000 – Request Reference FoI/24/270
Genetic Data

Information Requested:

How many individuals in Wales were identified to have a CACNA1C gene change as a result of genetic testing during the years 2021, 2022 and 2023?

Ideally, your response would indicate the total for each year separately with the classification of the finding (according to ACGS Guidelines i.e. Pathogenic, Likely pathogenic, Variant of Uncertain Significance, Likely benign, Benign and Conflicting interpretations of pathogenicity if you use this clinical significance group too) alongside the number of individuals identified.

In your answer, it would be helpful to know which diagnostic test method ie. narrow and specific panel (CACNA1C is listed in 4 separate cardiac panels in the Scottish Genomic Test Directory for Rare and Inherited Diseases) up to whole genome sequencing, was used to examine the potentially causative loci.

Response Details:

Please see the below table. Note, however, that the data provided is pulled from the auditable variant analysis databases of the All Wales Medical Genomics Service (AWMGS) that have been in use since approximately 2019. The AWMGS are unable to provide data where external laboratories have provided Welsh patients with sequencing results while also complying with the exemption outlined in section 12 of the Freedom of Information Act 2000. Also, the AWMGS may not hold full copies of results in its laboratory from external laboratories.

Year	2021 – WES	2022 – WES	2022 – WGS	2023 – WES	2023 – WGS
Benign	0	0	0	0	0
Likely Benign	0	0	0	0	0
VUS	0	3	1	0	0
Likely Pathogenic	0	0	0	0	0
Pathogenic	0	0	0	0	0

Key:

WES – whole exome sequencing with phenotype-specific gene panel.
WGS – whole genome sequencing.