

## Gene panel information

|                 |                                    |
|-----------------|------------------------------------|
| Gene panel      | Arrhythmia                         |
| Version         | 1                                  |
| Total genes     | 25                                 |
| Activation date | Friday 21 march 2025               |
| Publisher       | Center for Medical Genetics, Ghent |

## Genes

| Gene           | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes                                                                                                                                                                                                                                                                                                                                                                                                                      |
|----------------|--------------------------|--------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>CACNA1C</b> | 99.92 %                  | 114205       | Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3), Autosomal dominant                                                                                                                                  |
| <b>CALM1</b>   | 99.24 %                  | 114180       | Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3), Autosomal dominant; Long QT syndrome 14, 616247 (3), Autosomal dominant                                                                                                                                                                                                                                                                                       |
| <b>CALM2</b>   | 98.38 %                  | 114182       | Long QT syndrome 15, 616249 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                  |
| <b>CALM3</b>   | 99.9 %                   | 114183       | Long QT syndrome 16, 618782 (3), Autosomal dominant; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782 (3), Autosomal dominant                                                                                                                                                                                                                                                                                       |
| <b>CASQ2</b>   | 96.79 %                  | 114251       | Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive                                                                                                                                                                                                                                                                                                                                           |
| <b>CDH2</b>    | 99.82 %                  | 114020       | Arrhythmogenic right ventricular dysplasia 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant                                                                                                                                                                      |
| <b>DES</b>     | 99.93 %                  | 125660       | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Cardiomyopathy, dilated, 11, 604765 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant                                                                                                                                                                                                       |
| <b>DSC2</b>    | 99.51 %                  | 125645       | Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant                                                                                                                                                                            |
| <b>DSG2</b>    | 99.87 %                  | 125671       | Cardiomyopathy, dilated, 1BB, 612877 (3), Autosomal recessive; Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant                                                                                                                                                                                                                                                                                         |
| <b>DSP</b>     | 99.97 %                  | 125647       | Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive |
| <b>FLNC</b>    | 99.75 %                  | 102565       | Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia, familial, 617047 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant                                                                              |
| <b>JUP</b>     | 99.59 %                  | 173325       | Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                       |

# Arrhythmia

Gene panel

| Gene          | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |
|---------------|--------------------------|--------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>KCNH2</b>  | 99.79 %                  | 152427       | Short QT syndrome 1, 609620 (3); Long QT syndrome 2, 613688 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| <b>KCNJ2</b>  | 100 %                    | 600681       | Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| <b>KCNQ1</b>  | 99.9 %                   | 607542       | Short QT syndrome 2, 609621 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| <b>LMNA</b>   | 99.84 %                  | 150330       | Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant |
| <b>PKP2</b>   | 98.7 %                   | 602861       | Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| <b>PLN</b>    | 100 %                    | 172405       | Cardiomyopathy, dilated, 1P, 609909 (3); Cardiomyopathy, hypertrophic, 18, 613874 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| <b>RYR2</b>   | 99.77 %                  | 180902       | Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant; Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| <b>SCN5A</b>  | 99.86 %                  | 600163       | Ventricular fibrillation, familial, 1, 603829 (3); Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Long QT syndrome 3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; Brugada syndrome 1, 601144 (3), Autosomal dominant; Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive                                                                                                                                                                                                    |
| <b>SLC4A3</b> | 99.68 %                  | 106195       | Short QT syndrome 7, 620231 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| <b>TECRL</b>  | 99.8 %                   | 617242       | Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 (3), Autosomal recessive                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| <b>TGFB3</b>  | 99.97 %                  | 190230       | Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
| <b>TMEM43</b> | 99.95 %                  | 612048       | Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Auditory neuropathy, autosomal dominant 3, 619832 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
| <b>TRDN</b>   | 99.9 %                   | 603283       | Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 (3), Autosomal recessive                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |

## Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

\* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.