# UKGTN Testing Criteria

| **Test name:**<br>**Brugada Syndrome 6 Gene Panel** | |
|---|---|
| | |
| **Approved name and symbol of disorder/condition(s):**<br>See Appendix 1 | **OMIM number(s):** |
| **Approved name and symbol of gene(s):**<br>See Appendix 1 | **OMIM number(s):** |

| | |
|---|---|
| **Patient name:** | **Date of birth:** |
| **Patient postcode:** | **NHS number:** |
| **Name of referrer:** | |
| **Title/Position:** | **Lab ID:** |

**Referrals will only be accepted from one of the following:**

| **Referrer** | **Tick if this refers to you.** |
|---|---|
| Consultant Clinical Geneticist | |
| Consultant Cardiologist | |
| | |

**Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:**

| **Criteria** | **Tick if this patient meets criteria** |
|---|---|
| Index case with ECG consistent with Brugada syndrome **AND** | |
| A family history consistent with autosomal dominant inheritance **OR** | |
| A personal history of syncope without warning and/or aborted cardiac arrest. | |

**Additional Information:**

For panel tests:
At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation

**If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.**

## Appendix 1: Brugada Syndrome 6 Gene Panel

Genes in panel test and associated conditions
Rows that are highlighted in yellow show where the gene is currently being fully analysed in the context of a single separate UKGTN test

| HGNC standard name and symbol of the gene | HGNC number | OMIM number | OMIM standard name of condition and symbol | Mode of inheritance | OMIM number | Evidence of association between gene(s) and condition | % of horizontal coverage of gene | MLPA | Comments |
|---|---|---|---|---|---|---|---|---|---|
| *GPD1L* | HGNC:28956 | *611778 | Brugada syndrome 2 | AD | 611777 | PubMed: Various | 100% | No | Not available in UKGTN |
| *HCN4* | HGNC:16882 | *605206 | Brugada syndrome 8 | AD | 613123 | PubMed: 19165230 | 100% | No | Not available in UKGTN |
| *KCNE3* | HGNC:6243 | *604433 | Brugada syndrome 6 | AD | 613119 | PubMed: Various | 100% | No | Not available in UKGTN |
| *SCN1B* | HGNC:10586 | *600235 | Atrial fibrillation, familial, 13<br>Brugada syndrome 5 | AD<br>AD | 615377<br>612838 | PubMed: Various<br>PubMed: 18464934 | 98% | No | Not available in UKGTN |
| *SCN3B* | HGNC:20665 | *608214 | Brugada syndrome 7 | AD | 613120 | PubMed: 20031595 | 100% | No | Not available in UKGTN |
| *SCN5A* | HGNC:10593 | *600163 | Atrial fibrillation, familial, 10<br>Brugada syndrome 1<br>Cardiomyopathy, dilated, 1E<br>Long QT syndrome-3<br>Ventricular fibrillation, familial, 1 | AD<br>AD<br>AD<br>AD<br>AD | 614022<br>601144<br>601154<br>603830<br>603829 | PubMed: Various<br>PubMed: Various<br>PubMed: Various<br>PubMed: Various<br>PubMed: 10940383 | 100% | No | Available in UKGTN panel test |