Scottish Genomic Test Directory
New Format Guidance

You can navigate the test directory using the **filters in the column headers**. Select the **filter icon** to narrow your search by **disease group, indication, centre** and more.

	B	C	D	E	F	G	H	I	J	K	L
1	Disease Group	Indication	Referral Criteria	Requesting Specialties	Aberdeen	Dundee	Edinburgh	Glasgow	Test Method	Target/Genes	Routine Reporting Time
2	CARDIOLOGY	ANDERSEN-TAWIL SYNDROME	Ventricular arrhythmia and /or prolonged QTc; Periodic paralysis; Distinctive facial and skeletal features	Cardiologist with expertise in ICC, Clinical Genetics, Neurology, Paediatric Neurology	Y	N	N	N	Sanger	KCNJ2, KCNJ5	56
3	CARDIOLOGY	ARRHYTHMIA PANEL	Out of Hospital Cardiac Arrest with no known cause; Sudden cardiac death with negative post mortem	Cardiologist with expertise in ICC, Clinical Genetics, Pathology in discussion with Clinical Genetics	Y	N	N	N	NGS	KCNQ1*, KCNH2*	112
4	CARDIOLOGY	arrhythmogenic right ventricular cardiomyopathy	A possible, borderline, or definite diagnosis according to 2010 modified Task Force criteria; Fibrosis & fatty replacement of myocardium affecting one or both ventricles seen on Echocardiogram or Post-mortem investigations; Clinical phenotype considered to be compatible with ACM (e.g., dilated cardiomyopathy, arrhythmia, heart failure)	Cardiologist with expertise in ICC, Clinical Genetics, Pathology	Y	N	N	N	NGS	PKP2, DSG2, DSC	112
5	CARDIOLOGY	ATRIAL FIBRILLATION	Atrial fibrillation detected at young age with family history of atrial fibrillation or sudden cardiac death	Cardiologist with expertise in ICC, Clinical Genetics	Y	N	N	N	NGS	SCN5A, ABCC9, G	56
6	CARDIOLOGY	BARTH SYNDROME	Cardiomyopathy; Neutropenia; Fatigue & general muscle weakness; Growth/feeding issues	Cardiology, Clinical Genetics, Paediatrics	Y	N	N	N	Sanger	TAFAZZIN (TAZ)	56
7	CARDIOLOGY	BRUGADA SYNDROME AND SODIUM CHANNEL DISEASE	Cardiac arrest in the absence of secondary causes, most commonly at night; Arrhythmia triggered by fever; Type 1 Brugada ECG; Atrial arrhythmia, sinus node dysfunction, or conduction disease, with or without QT prolongation predominantly in children and young people	Cardiologist with expertise in ICC, Clinical Genetics	Y	N	N	N	NGS	SCN5A, CACNA1C	112
8	CARDIOLOGY	CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)	Ventricular fibrillation or polymorphic VT; Bi-directional VT on exercise; Resuscitated from cardiac arrest, or syncope compatible with tachyarrhythmia especially related to physical activity, or acute emotion, in the presence of an unremarkable ECG (e.g. normal QT interval), and in the absence of structural heart or coronary artery disease; Family history of premature sudden cardiac death particularly due to physical activity or emotion	Cardiologist with expertise in ICC; Clinical Genetics; Pathology in discussion with Clinical Genetics	Y	N	N	N	NGS	RYR2, CALM1, CALM2, CASQ2, DPP6, TRDN	56

Summary | RID TD V6 | Solid Tumour TD V6 | Haemonc TD V6 | Pharmacogenomics | Change Summary V6

The **Summary** tab provides an introduction and summary of key aspects of the test directory including, laboratory contact information, sample requirements and clinical consent implications.

Navigate between the **Rare and Inherited Disease (RID), Solid Tumour, Haematological-Oncology (Haemonc) and Pharmacogenomic** test directories using these tabs.

The **Change Summary** tab highlights the updates made since the last version of the test directory.