

## 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.

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4	В	С	D	Е		F	G	Н	- 1	J	K	L
					Т							
1 Dise	ase Group	ndication	Referral Criteria	Requesting Specialties	Ψ Δ	herdeen 🔻	Dundee V	Edinburgh V	Glasgow V	Test Method 🔻	Target/Genes ▼	Routine Reporting Time 🔻
1 0100	азо отоар	naidation	Hororiat Gritoria	noquesting operations		шониооп	Danaco	Lambargii	otasgon	rostriotilou	raigod ochos	modeline neporting rime
2 CARI	DIOLOGY	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	V	,	N	N	N	Sanger	KCNJ2, KCNJ5	56
2 Criti	3102001	AND ENDER TANTE OTHER DROWLE	pararysis, bistilicave racial and skeretar reatures	deneties, rearringy, racardine rearringy	+			<u>"</u>		ounger	Konsz, Konss	30
			Out of Heavital Conding Asset with an Issues access Sudden	Cardiologist with expertise in ICC, Clinical								
3 CARI	DIOLOGY	ARRHYTHMIA PANEL	Out of Hospital Cardiac Arrest with no known cause; Sudden cardiac death with negative post mortem	Genetics, Pathology in discussion with Clinical Genetics	Y	,	N	N	N	NGS	KCNQ1*, KCNH2*	112
			A possible, borderline, or definite diagnosis according to 2010		$\top$							
			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									
		arrhythmogenic right ventricular	compatible with ACM (e.g., dilated cardiomyopathy, arrhythmia, heart	Cardiologist with expertise in ICC, Clinical Genetics,								
4 CAR	DIOLOGY	cardiomyopathy	failure)	Pathology	Υ	,	N	N	N	NGS	PKP2, DSG2, DSC	112
			Atrial fibrillation detected at young age with family history of atrial									
5 CAR	DIOLOGY	ATRIAL FIBRILLATION	fibrillation or sudden cardiac death	Cardiologist with expertise in ICC, Clinical Genetics	Υ	,	N	N	N	NGS	SCN5A, ABCC9, G	56
6 CAR	DIOLOGY	BARTH SYNDROME	Cardiomyopathy; Neutropenia; Fatigue & general muscle weakness; Growth/feeding issues	Cardiology, Clinical Genetics, Paediatrics	l,	,	N	N	N	Candar	TAFAZZIN (TAZ)	56
0 CAN	DIOLOGI	BANTHSTNDNOME	Growth/reeding issues	Cardiology, Clinical Genetics, Faediatrics	+		IN	IN	IN	Sanger	TAFAZZIN (TAZ)	30 .
			Cardiac arrest in the absence of secondary causes, most commonly at									
		DDUOADA OVALDDOME AND	night; Arrhythmia triggered by fever; Type 1 Brugada ECG; Atrial									
7 CAR	DIOLOGY	BRUGADA SYNDROME AND SODIUM CHANNEL DISEASE	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	V	,	N	N	N	NGS	SCN5A, CACNA1	112
7 07111	5.02501	S S S S S S S S S S S S S S S S S S S	Without Q1 protongation prodominantly in orintaren and young people	our are to great with expertise in ree, earned echecies	+			<u>"</u>			0011011, 071011712	
			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									
		CATECHOLAMINERGIC	interval), and in the absence of structural heart or coronary artery								RYR2, CALM1,	
		POLYMORPHIC VENTRICULAR	disease; Family history of premature sudden cardiac death particularly								CALM2, CASQ2,	
8   CAR	DIOLOGY	TACHYCARDIA (CDVT)	due to physical activity or emotion	Patholog, in discussion with Clinical Genetics	Y	•	N .	N	IN	NGS	DPP6, TRDN	56 :
<	>	Summary RID TD V6 Soli	d Tumour TD V6 Haemonc TD V6 Pharmacogenomic	Change Summary V6 +				1				

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.