

CPVT patient pathway

Clinical presentation	GP DGH	Referral Point	Investigations	Phenotype	Genetic testing	Management	Follow-up
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Index case	Refer Urgent if syncopal or unexplained seizures	ICC Clinic	FH, ECG, Tape, ExECG +/- epinephrine test	Probable or definite CPVT	YES – RYR2, CASQ2, Consider KCNJ2, Calm, Ank, LQT 1,4,5. TRD Channelopathy panel	Lifestyle advice and consider SCD risk	YES - evaluate first degree relatives
			Exclude metabolic and structural disease as causes **	Low likelihood	NO	Management appropriate to any underlying condition – discharge otherwise	

Relative (first degree blood relative) with CPVT (Disease mutation may be known)	Refer Urgent if syncopal or unexplained seizures	ICC Clinic	Disease mutation identified in family – gene test & counsel: <ul style="list-style-type: none"> Asymptomatic- ECG and ExECG +/- epinephrine test Symptomatic - ECG, Tape, ExECG +/- epinephrine test 	NB Ensure SCD risk assessed especially if symptomatic Cascade genetic test –ve – discharge and manage any symptoms appropriately Cascade genetic test +ve continue pathway at †			
			VUS identified or Disease mutation absent in family	Probable or definite CPVT	YES - if family untested YES - if VUS present to inform segregation study NO - if mutation absent	Lifestyle advice and consider SCD risk	YES - evaluate family according to pedigree
			FH +/- segregation study, ECG, Tape, ExECG +/- epinephrine test	Low likelihood	NO	Lifestyle advice only (precautionary)	Dependent on age and sex

More distant blood relative: Seek advice from ICC clinic. Refer key relatives who are likely carriers and/or suffer unexplained syncope or seizures

** Consider K/Mg/Ca and thyroid function, echocardiography and cardiac MRI to exclude ARVC and coronary angiogram in older patient
VUS = variant of unknown significance – pathogenic effect can be assessed by segregation study of variant and phenotype, FH = family history

† Relative with positive genetic test	ICC Clinic	Family history, Tape, ExECG	Lifestyle advice and consider SCD risk	YES - evaluate family according to pedigree
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