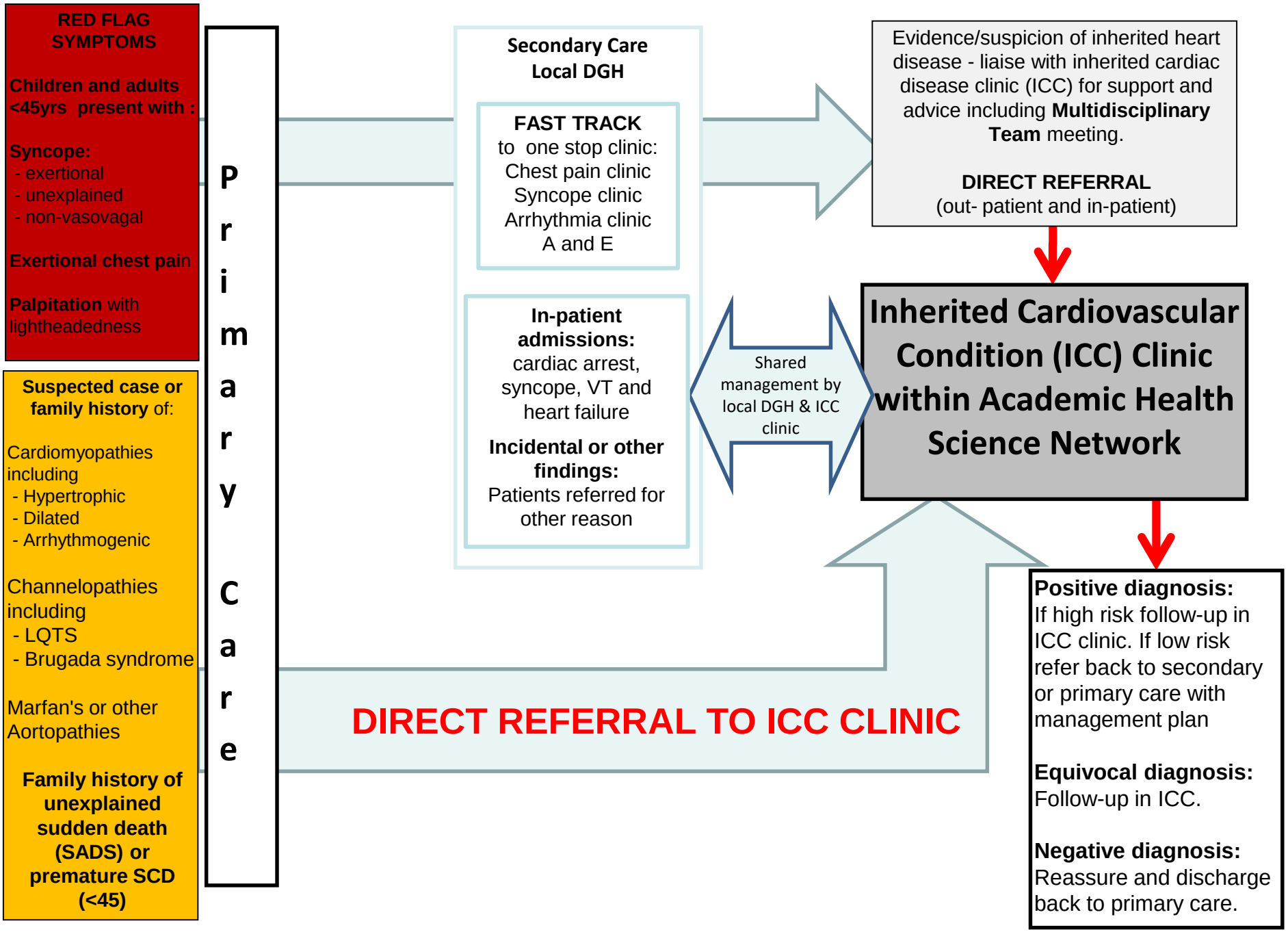
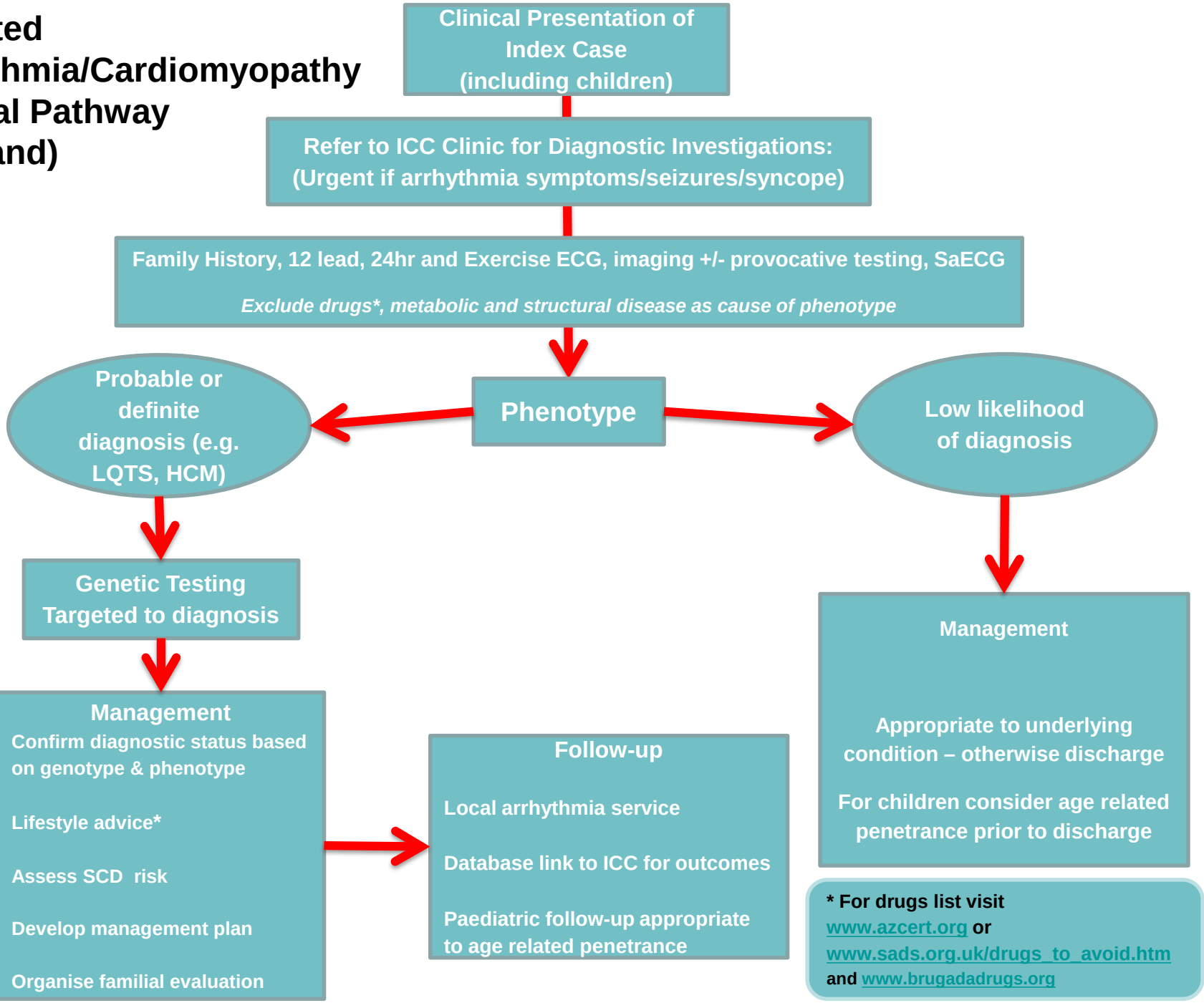
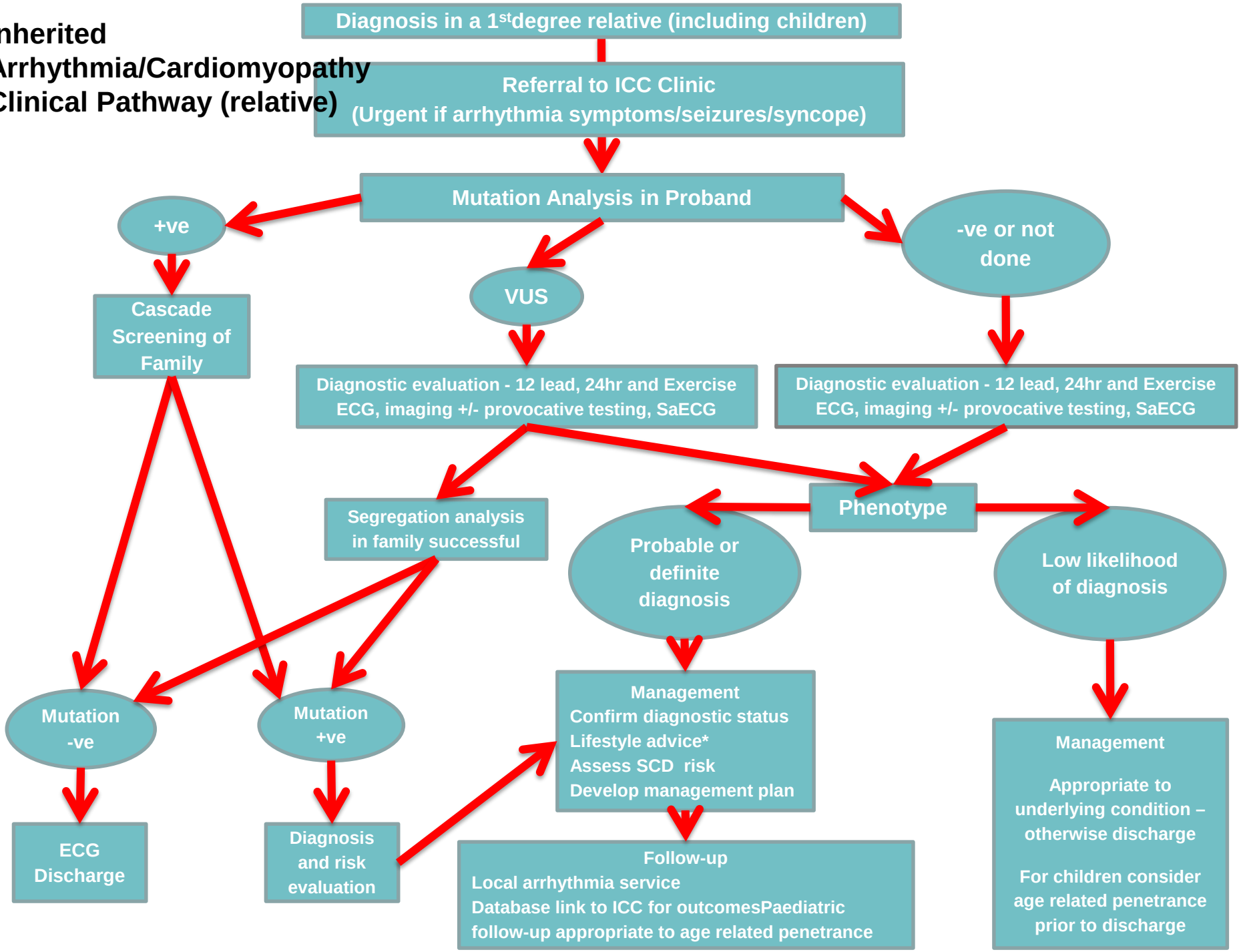

RED FLAG SYMPTOMS
Children and adults <45yrs present with :
Syncope:
- exertional
- unexplained
- non-vasovagal
Exertional chest pain
Palpitation with lightheadedness
Suspected case or family history of:
Cardiomyopathies including
- Hypertrophic
- Dilated
- Arrhythmogenic
Channelopathies including
- LQTS
- Brugada syndrome
Marfan's or other Aortopathies
Family history of unexplained sudden death (SADS) or premature SCD (<45)
Primary Care
Secondary Care Local DGH
FAST TRACK to one stop clinic:
Chest pain clinic
Syncope clinic
Arrhythmia clinic
A and E
In-patient admissions:
cardiac arrest, syncope, VT and heart failure
Incidental or other findings:
Patients referred for other reason
Shared management by local DGH & ICC clinic
Evidence/suspicion of inherited heart disease - liaise with inherited cardiac disease clinic (ICC) for support and advice including Multidisciplinary Team meeting.
DIRECT REFERRAL
(out- patient and in-patient)
Inherited Cardiovascular Condition (ICC) Clinic within Academic Health Science Network
DIRECT REFERRAL TO ICC CLINIC
Positive diagnosis:
If high risk follow-up in ICC clinic. If low risk refer back to secondary or primary care with management plan
Equivocal diagnosis:
Follow-up in ICC.
Negative diagnosis:
Reassure and discharge back to primary care.

Inherited Arrhythmia/Cardiomyopathy Clinical Pathway (proband)

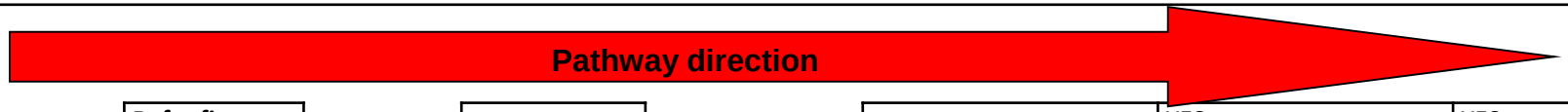


Inherited Arrhythmia/Cardiomyopathy Clinical Pathway (relative)



SADS families: Patient Management Pathway

Phenotype	GP	Referral Point	Investigations	Phenotype identified in relative?	Genetic testing	Follow-up	Therapy?
SADS: Unexplained SCD (also consider Sudden Unexplained Death in epilepsy [SUDEP] as SADS until proven otherwise) Investigations before referral: Expert autopsy Retention of fresh tissue or blood for freezing +/- DNA extraction	**Refer first degree blood relatives including children** Unexplained syncope or seizures? Refer urgently	**ICC clinic**	Expert pathology Review coronial reports, PMH, old ECGs Investigations can include: Family history, ECG, Echo, ExECG +/- SaECG, Tape and Ajmaline test Children to complete investigation as age permits	**YES**	YES Appropriate to phenotype (see recommended pathway) Consider role for a molecular autopsy of SADS victim if suitable tissue retained	YES Appropriate to phenotype and refer at risk relatives	YES Appropriate to phenotype and risk
	More distant blood relative: Seek advice from ICC clinic Refer key relatives who are likely carriers and/or suffer unexplained syncope or seizures			**NO**	Consider de novo disease and role for a molecular autopsy of SADS victim if suitable tissue retained	Follow-up children when able to complete testing and then once during, once after puberty and then once in adulthood, investigating as appropriate. Discharge adults Re-refer: • urgently if symptomatic • if a phenotype is identified in another blood relative	NO



Clinical presentation	GP DGH	Referral Point	Investigations	Phenotype	Genetic testing	Management	Follow-up
	Pathway direction						
Index case	Refer **Urgent if syncopal or unexplained seizures**	**ICC Clinic**	FH, ECG, Tape, ExECG +/- epinephrine test Exclude drugs*, metabolic and structural disease as causes **	Probable or definite LQTS	YES – KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2	Lifestyle advice* and consider SCD risk	YES - evaluate first degree relatives
				Low likelihood	NO	Management appropriate to any underlying condition – discharge otherwise	
Relative with FH of LQTS in a blood relative (Disease mutation may be known)	Refer **Urgent if syncopal or unexplained seizures**	**ICC Clinic**	Disease mutation identified in family – gene test & counsel: • Asymptomatic relative ECG • Symptomatic relative 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 FH +/- segregation study, ECG, Tape, ExECG +/- epinephrine test	Probable or definite LQTS	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
				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

* For drugs list visit www.azcert.org or www.sads.org.uk/drugs_to_avoid.htm

** Consider K/Mg/Ca and thyroid function, echocardiography and cardiac MRI

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

Long QT Syndrome:Patient Management Pathway

Clinical presentation	GP DGH	Referral Point	Investigations	Phenotype	Genetic testing	Management	Follow-up
			Pathway direction				
Index case	Refer **Urgent if syncopal or unexplained seizures**	**ICC Clinic**	FH, ECG*, Tape, +/- SaECG, ExECG, ajmaline test * Exclude drugs, metabolic and structural disease as causes **	Brugada syndrome: Brugada type 1 ECG pattern present spontaneously or with ajmaline	May be useful – SCN5A +/- other associated genes	Lifestyle advice ** and stratify SCD risk	YES - evaluate first degree relatives
				Brugada type 1 ECG pattern absent even after ajmaline test	NO	Management appropriate to any underlying condition – discharge otherwise	
Relative with FH of Brugada syndrome in a blood relative (Disease mutation may be known)	Refer **Urgent if syncopal or unexplained seizures**	**ICC Clinic**	Disease mutation identified in family ECG +/- ajmaline test*	Ensure SCD risk assessed especially if symptomatic Cascade genetic test +ve – continue pathway at † Cascade genetic test –ve – discharge if disease mutation of high penetrance and ECG +/- ajmaline test –ve			
			Family untested or mutation absent FH +/-segregation study, ECG, SaECG, ExECG, Tape +/- ajmaline test*	Brugada type 1 ECG pattern	May be useful if family untested NO - if mutation absent	Lifestyle advice** and consider SCD risk	YES - evaluate family according to pedigree
				Brugada type 1 ECG pattern absent even after ajmaline	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			* ECG and ajmaline testing should utilise high right ventricular leads in patients without a resting type 1 ECG pattern. Tape ideally should have right ventricular leads. Flecainide can replace ajmaline but is not ideal due to lower sensitivity. ** For drug list see www.brugadadrugs.org. Consider K, Mg, thyroid function, echocardiography, cardiac MRI and coronary angiography.				
† Individual with positive cascade genetic test		**ICC Clinic**	ECG, Tape +/- ajmaline test*, SaECG			Lifestyle advice** and risk stratify for SCD	YES - evaluate family according to pedigree

Brugada Syndrome:Patient Management Pathway