Inherited cardiac conditions are common but often misdiagnosed so it is essential health professionals know the symptoms and when to refer to cardiac experts.

**Treatment of inherited cardiac conditions**

**In this article...**
- The pathology of inherited cardiac conditions
- Definition and UK prevalence of inherited cardiomyopathies
- Definition and UK prevalence of inherited arrhythmias

**KEY POINTS**

1. An estimated 12 people aged 35 years or under die each week in the UK from undiagnosed cardiac conditions.

2. Most sudden cardiac deaths are as a result of inherited cardiomyopathies and arrhythmias.

3. Many patients with cardiomyopathies and arrhythmias are undiagnosed or their symptoms are thought to be due to another illness.

4. Difficulties in diagnosing arrhythmias make it almost impossible to clinically screen relatives.

5. Treatment of cardiomyopathy will not cure the patient, but can control symptoms.

**Resources**

- Association for Inherited Cardiac Conditions: www.aicc-uk.co.uk
- The British Heart Foundation: www.bhf.org.uk
- The Cardiomyopathy Association: www.cardiomyopathy.org
- Cardiac Risk in the Young: www.c-r-y.org.uk
- Rare Disease UK: www.raredisease.org.uk
- Sudden Arrhythmia Death Syndrome: www.sads.org

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**Authors**

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**Abstract**


Inherited cardiac conditions are difficult to detect and screen for. Part 2 of our six part series on rare diseases explains why health professionals need to be aware of the symptoms associated with inherited cardiac conditions so they can make swift referrals to expert services.

It is estimated that every week in the UK, 12 apparently fit and healthy young people aged 35 years or under die from undiagnosed cardiac conditions (Papadakis et al, 2009). Sudden cardiac death (SCD) or a serious event in an otherwise healthy young person raises the possibility of an underlying genetic condition and suggests their relatives may also be at risk.

The underlying cardiac pathology of inherited cardiac conditions (ICCs) can be structural or non-structural and fall into five main categories:
- Cardiomyopathies;
- Arrhythmias;
- Aortopathies;
- Muscular dystrophies;
- Familial hypercholesterolaemia.

The majority of SCDs are due to inherited cardiomyopathies and arrhythmias (Elliott et al, 2011). Genetic diseases of the heart are caused by a mutation in one or more of the genes that affect the structure or function of the heart muscle or conduction pathways (Kumar and Elliott, 2010).

The majority of ICCs are inherited in dominant genes, with a small number inherited recessively.

**Conditions**

Cardiomyopathies can be divided into five specific subgroups: hypertrophic; dilated; arrhythmogenic; restrictive; and unclassified (Elliott et al, 2008). Table 1 gives the definitions and prevalence of these subgroups.

The clinical course of cardiomyopathies and arrhythmias is variable. Many patients are asymptomatic or present with symptoms that can be misdiagnosed as asthma or epilepsy. Symptoms can include:
- Shortness of breath (dyspnoea);
- Chest pain (angina);
- Irregular heartbeat (palpitations);
» Light-headedness or dizziness (presyncope);
» Fainting (syncope);
» Sudden unexplained cardiac death.

**Arrhythmias/cardiac ion channelopathies**

**Diagnosis**

Long QT syndrome, short QT syndrome and Brugada syndrome belong to a group of arrhythmic syndromes called cardiac ion channelopathies (CICs) (Martin et al, 2012). Other rare arrhythmia syndromes include catecholaminergic polymorphic ventricular tachycardia and Wolff-Parkinson-White syndrome (Table 2).

An arrhythmia that causes SCD leaves no signs and can be virtually impossible to diagnose during a post mortem, even by an expert cardiac pathologist. This makes it difficult to effectively clinically screen first-degree relatives and requires the expertise of a clinician with appropriate CIC expertise and advanced electrocardiogram interpretation skills, such as a consultant cardiologist or electrophysiologist (Cardiac Risk in the Young, 2003). Symptom onset is often sudden, and includes palpitations, presyncope or syncope.

It is important to recognise patients who may be presenting with an arrhythmic syndrome or CIC and to seek prompt medical assessment from an expert clinician (Wilson et al, 2008). As many of these syndromes are likely to be inherited, it may be appropriate to refer immediate family members quickly but sensitively for clinical screening and genetic testing. Several regional ICC services across the country provide counselling, clinical screening and genetic testing; a list of these is on the Association for Inherited Cardiac Conditions website (Box 1).

**Treatment**

Treatment for cardiomyopathies and arrhythmias will not reverse the condition but is often effective in controlling symptoms, particularly if used in conjunction with lifestyle changes.

If a patient’s risk stratification shows a risk of SCD, the cardiologist will discuss the need for an implantable cardiac defibrillator (CRY, 2003).

Sometimes, cardiomyopathy symptoms can mimic those of congestive heart failure (especially activity intolerance and

### Table 1. Prevalence and Definitions of UK Inherited Cardiomyopathies

<table>
<thead>
<tr>
<th>Condition</th>
<th>UK population prevalence</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>1:500</td>
<td>Defined by the presence of increased ventricular wall thickness or mass without a loading condition such as hypertension or valvular disease (Elliott, 2008)</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>1:2500</td>
<td>Characterised by dilatation of the cardiac chambers with impaired contraction of the ventricles (Burkett and Hershberger, 2005)</td>
</tr>
<tr>
<td>Arrhythmogenic right ventricular cardiomyopathy</td>
<td>1:1000 to 1:10,000</td>
<td>Dilated, poorly functioning right ventricle with fatty deposits within the walls. Patients are at risk of abnormally fast, life-threatening heart rhythms (ventricular tachycardia) (Sen-Chowdhry et al, 2004)</td>
</tr>
<tr>
<td>Restrictive cardiomyopathy</td>
<td>1:1000 to 1:5000</td>
<td>Restricted ventricular filling, typically biatrial enlargement with reduced diastolic volume of either or both ventricles (Kumar and Elliott, 2010)</td>
</tr>
<tr>
<td>Left ventricular non-compaction cardiomyopathy</td>
<td>0.12 cases per 100,000</td>
<td>An anatomic abnormality of left ventricular myocardial development, leaving deep, almost sponge-like trabeculations in the left ventricle myocardium (<a href="http://www.cardiomyopathy.org/Left-ventricular-noncompaction.html">http://www.cardiomyopathy.org/Left-ventricular-noncompaction.html</a>)</td>
</tr>
<tr>
<td>Takotsubo cardiomyopathy</td>
<td></td>
<td>Also referred to as apical ballooning syndrome and stress cardiomyopathy as it can be caused by extreme stress (Sharkey et al, 2010). Temporary condition where the heart muscle becomes suddenly weakened</td>
</tr>
</tbody>
</table>

### Table 2. Prevalence and Definitions of UK Inherited Arrhythmias

<table>
<thead>
<tr>
<th>Condition</th>
<th>UK population prevalence</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Long QT syndrome</td>
<td>1: 2500</td>
<td>The most common of the ion channelopathies, characterised by prolonged ventricular repolarisation and QT interval (corrected for heart rate) (Maron et al, 2006)</td>
</tr>
<tr>
<td>Short QT syndrome</td>
<td>100 published cases since 2000</td>
<td>Characterised by a short QT interval (&lt;330ms) on an ECG with tall peaked T-waves (Bjernegaard and Gussak, 2013)</td>
</tr>
<tr>
<td>Brugada syndrome</td>
<td>1: 5000</td>
<td>Has a distinctive ECG that is often concealed until provoked by an external influence such as certain drugs, for example ajmaline or flecainide (Maron et al, 2006)</td>
</tr>
<tr>
<td>Catecholamine polymorphic ventricular tachycardia</td>
<td>1: 10,000</td>
<td>Often triggered by acute emotion or extreme physical exertion shows as a bidirectional ventricular tachycardia (Maron et al, 2006)</td>
</tr>
<tr>
<td>Familial Wolff-Parkinson-White Syndrome</td>
<td>1.5-31: 1000</td>
<td>The second most common cause of paroxysmal supraventricular tachycardia due to conduction through an accessory pathway (Gollob et al, 2001)</td>
</tr>
</tbody>
</table>

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dyspnoea). It is essential to get an accurate diagnosis as treatment is very different - diuretics work well in heart failure but can exacerbate symptoms in cardiomyopathy (Jessop et al, 2009).

Beta-blockers (such as nadolol or bisoprolol) are commonly used for both cardiomyopathies and arrhythmias. These can slow the heart rate and make the heart contract less forcefully, allowing more time for the ventricle to fill with each heartbeat and helping to reduce chest pain, breathlessness and palpitations. Competitive sports are not advisable for patients taking beta-blockers due to the possibility of sudden collapse or increased heart failure (Gersh et al, 2011).

The role of clinical nurse specialists is to facilitate streamlined care for referred families. Nurses often feel frightened and unsure of the next steps – as specialist nurses we “walk alongside” them through the process. We offer support and guidance, obtain the family history, organise cardiac investigations and ensure relevant medical notes are available to coordinate families through a one-stop shop clinic with cardiologists and geneticists for clinical screening, genetic testing and follow-up where needed. NT

References

Nurses’ role
The role of clinical nurse specialists is to facilitate streamlined care for referred families.

We are often the first point of contact for those who have lost a relative due to SCD or are diagnosed with an ICC that may affect other family members. Families often feel frightened and unsure of the next steps – as specialist nurses we “walk alongside” them through the process. We offer support and guidance, obtain the family history, organise cardiac investigations and ensure relevant medical notes are available to coordinate families through a one-stop shop clinic with cardiologists and geneticists for clinical screening, genetic testing and follow-up where needed. NT

Leo

Could you be my parents?

All about me...

“Leo is a lovely boy with a delightful smile. He was placed with local authority carers following his birth. Leo appears to be thriving and meeting all his developmental milestones, he is starting to say a few words like “ta”, “yes”, “bath time” etc...

He is walking independently and is an active boy. Leo attends a baby gym three times a week and is reported to enjoy the interaction with other children.

Leo has inherited an extremely rare disorder called congenital central hypoventilation syndrome (CCHS), like his mum. This condition involves a failure of the breathing centres in the brain stem to respond to the body’s needs to take in oxygen and get rid of carbon dioxide whilst asleep. Leo’s life expectancy is likely to be of normal duration, provided he always receives assisted ventilation when he is asleep and undergoes periodic reviews for his condition.

We are looking for a two parent family for Leo, who can provide a secure, loving, permanent home for him.

To find out more about Leo please contact:

Yasmeen Akhtar, Family Placements Team, on 0845 352 8609
Email: yasmeen.akhtar@sandwell.gov.uk

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