Assessing Risk in Hypertrophic Cardiomyopathy

Prognosis in patients with hypertrophic cardiomyopathy (HCM) is generally good but a small subgroup of patients die suddenly because of abnormally fast heart rhythms called ventricular arrhythmias.

Once a ventricular arrhythmia becomes sustained (longer lasting), the heart is unable to pump normally, rapidly leading to death. A smaller proportion of sudden deaths are probably caused by abnormally slow heart rhythms.

The annual risk of sudden cardiac death (SCD) in contemporary medical literature is less than one per cent per year and fortunately only a small proportion of HCM patients die suddenly. Despite intense research in this field, SCD remains a complex and incompletely understood clinical problem.

Prevention of SCD

Avoiding Exercise

Intense exertion may bring on ventricular arrhythmias and SCD. International guidelines have historically recommended that patients with HCM avoid competitive sports and strenuous physical effort. Even though the effectiveness of this approach is unclear, on balance the medical community feels that this is a reasonable recommendation. Participation in high/moderate intensity exercise, if desired, can be considered in some patients following an expert assessment.

Drugs

Drugs which help control heart rhythm (such as amiodarone and sotalol) do not provide adequate protection from SCD and should not be relied on for SCD prevention. These drugs are still used in HCM patients as they are effective in controlling other heart rhythm abnormalities such as atrial fibrillation.

Implantable cardioverter defibrillators (ICDs)

These devices, similar to pacemakers, monitor each heart beat and deliver an electric shock if a potentially life threatening ventricular arrhythmia develops.

An ICD shock stops a ventricular arrhythmia and restores sinus rhythm. Implanted devices are currently the most effective means of preventing SCD. Unfortunately ICDs have drawbacks:

- Implanting an ICD involves an operation that carries a small risk, in some cases, of causing damaging the heart or lungs;
- The battery will eventually deplete, so replacement is needed around every five to ten years;
- Implantation of an ICD can in some cases result in an infection at the site of insertion, which in some cases can affect the leads;
- ICD hardware malfunction is very rare but in some cases may require removal of the device;
- ICDs can occasionally and in certain circumstances deliver a therapy shock to a rhythm which isn't a dangerous rhythm (the ICD can be reprogrammed to reduce the likelihood of this happening); and
- Lifestyle modifications may need to be implemented (e.g. advice around contact sports) and drivers should inform the DVLA as some restrictions apply depending on the type of license.

All these complications are rarely life threatening and the ICD is unequivocally the most successful treatment for preventing SCD in HCM.

Deciding on ICD treatment

When deciding whether an ICD should be implanted, patients and their medical team need to consider:

- The risk of sudden death and the implications of no treatment; and
- The benefits and the risks of treatment with an ICD.

Specialist nurses and doctors should provide adequate information to allow patients to reach an informed decision in line with shared decision making based on real world data, as well as individual preferences.

The perception of risk does vary between patients and their clinical team, and the threshold for treatment may depend on non-medical circumstances such as having a young family or lifestyle modification after ICD implantation. Discussion with other patients who already have an ICD may be helpful when considering treatment.

Assessing the risk of SCD

Ascertaining the magnitude of SCD risk is essential so that patients at high enough risk, who would benefit most from an ICD, can be identified. All HCM patients should have a comprehensive assessment.

In the small number of people who survive a cardiac arrest there is a high risk of recurrence. Clinicians agree ICDs are beneficial in these patients.

The vast majority of HCM patients do not have a prior

episode. The risk assessment involves a clinical review, an ultrasound scan of the heart (echocardiogram), and a heart rhythm monitor over 24 to 48-hours. On occasions an exercise test on a bicycle or treadmill can be helpful as well as a cardiac magnetic resonance image (cMRI).

The risk of SCD should then be reassessed every one to two years, or if there are changes or new symptoms, such as fainting.

Guidance by the AHA/ACC 2020 and the 2023 European guidelines, advise assessment of risk factors associated with SCD, for example:

- Unexplained collapse with loss of consciousness i.e.an episode of fainting with no obvious medical explanation. Fainting after having blood taken or because of a slow heart rate is not considered a marker of high risk. A sudden faint with no warning is more suspicious.
- A history of SCD in a first degree relative (parent, sibling or child). A family tree will be drawn up and relatives who died suddenly at a young age identified. This is sometimes limited by a lack of information.
- Short episodes of ventricular arrhythmia during heart rhythm monitoring. Most patients are not aware of these but they are a marker of risk, particularly in a young patient or during exercise.
- Extreme thickening of the main pumping chamber of the heart seen in an echocardiogram. Sometimes a magnetic resonance scan (MRI) of the heart may be needed.

With a few exceptions, none of these risk factors are strong enough to predict SCD on their own.
To overcome this problem, guidelines recommend implantation of ICDs in patients with multiple risk factors (such as family history of SCD as well as unexplained fainting) or in some patients with a single risk factor.

This approach has been in clinical use since 2003 and is partially successful. Research which critically appraised the robustness of this approach demonstrated several limitations, which lead to the development of a new method for risk stratification.

HCM Risk-SCD

To overcome the limitations above, a collaboration of European investigators developed a method of assessing the risk of SCD in 2013. This is called HCM Risk-SCD and since its publication has been endorsed by the 2023 European Society of Cardiology guidelines and the AHA/ACC 2020 guidelines as the advised method for risk stratification.
As well as the risk factors listed above, age, the size of the left atrium of the heart and obstruction to the blood flow out of the heart are also considered to estimate the risk of SCD within five years.

This model provides a numerical estimate of risk as a percentage. A young HCM patient with unexplained fainting has a higher risk of SCD than an older patient without fainting. The five year estimate of SCD risk can be used by patients and their clinical team to consider whether the risks of having an ICD outweigh the benefits.

Additional risk factors can be considered to supplement HCM Risk-SCD estimations e.g. the degree of scarring in the heart muscle (this is determined by a CMR scan) and declining heart function.

HCM Risk-SCD can be used in most patients with HCM. However, in patients with Anderson-Fabry disease or other rarer causes of HCM, this method should not be used. It is also not clear how to use it in patients who have had treatment for obstruction (an alcohol septal ablation or myectomy) after their initial assessment. Patients with a history of cardiac arrest should receive an ICD irrespective of their HCM Risk-SCD score.

Conclusions and future directions

Predicting the risk of SCD, irrespective of method or heart condition, is challenging. HCM Risk-SCD is now the standard of care, but it is not perfect and currently it is not possible to predict all cases of SCD.

Improvements in ICD technology will also help reduce ICD complications and help tip the balance in favour of treatment in some patients.

Further information:

To learn more about myectomy and septal ablation, visit: **https://youtu.be/h4LL37rgAhM**

To learn more about driving and cardiomyopathy, visit: www.cardiomyopathy.org/driving

To learn more about how we can support you, visit: www.cardiomyopathy.org/support

Thank you to Dr Costas O'Mahony, St Bartholomew's Hospital, London for his input on this resource

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