Commissioning Guide: Services for patients with inherited cardiovascular conditions

Key messages

1. Inherited cardiovascular conditions may result in sudden cardiac death if undiagnosed or not managed with sufficient expert clinical input.

2. Care for this group of patients needs to be delivered by a multi-disciplinary team which includes cardiology, pathology and genetics expertise.

3. Inherited cardiovascular conditions affect families and since most are inherited in an autosomal dominant pattern, 50% of the index patient’s relatives may have inherited the condition.

4. Through genetic testing, appropriate use of devices such as ICDs and targeted use of drugs and preventive strategies according to the genetic basis of the condition, care can be targeted to those at risk and many deaths could be prevented.

5. Currently there is inequity in service provision across the UK with commonly 10-20 fold variation in levels of service provision.

6. London and SE Coast regional figures can be used as a guide for information on estimated regional service provision.
Context

Inherited cardiovascular conditions cause sudden cardiac death in many apparently healthy young people and adults in the UK every year. Many of these deaths could be prevented if timely diagnosis and expert management of these patients was in place across the UK. New knowledge on the molecular, particularly the genetic basis of these conditions, could enable the detection of these conditions in individuals who do not yet display symptoms or who have early signs of the conditions. Use of genetic tests, as opposed to conventional clinical surveillance, can reduce costs as family members who do not carry the mutation can be discharged from follow-up. It may not be possible to prevent all sudden cardiac deaths caused by inherited disease, but many families could be spared the trauma of multiple deaths in family members who share the same genetic mutations underlying the disease. Chapter 8 of the National Service Framework for Coronary Heart Disease covers arrhythmias and focuses attention on preventing sudden cardiac death where possible.

In the current environment, it is vital that local health economies work with the system to focus on quality and productivity and collectively, we need to ensure we provide the tools for leaders to do so locally. Organisations across the system will need to work together to identify and prioritise activities which are safe, offer value for money and improve quality and the patient experience. This document, with its component parts, will help commissioners of inherited cardiovascular conditions services to commission quality services.

Recent innovations in the diagnosis and management of individuals with inherited cardiovascular conditions have the potential to improve health outcomes for affected individuals and to save lives of family members through cascade clinical assessment and genetic testing. However, there is no specific NICE, HTA or other evidence based guidance related to this group of disorders, although recent NICE guidance has been published on the identification and management of familial hypercholesterolaemia¹ (National Institute for Health and Clinical Excellence 2008). This Commissioning Guide is based on the Report of a working group of experts working in the field of inherited cardiovascular conditions (ICCs) entitled Heart to Heart Inherited Cardiovascular Conditions Services: A Needs Assessment and Service Review² and on evidence from patient and voluntary organisations and a sub-group of those with expertise on the ethical, legal and social issues associated with ICCs.

Although many of the individual conditions are uncommon, collectively the conditions represent a substantial burden of disease with approximately 340,000 people having an inherited cardiovascular condition in the UK (see Heart to Heart Inherited Cardiovascular Conditions Services: A Needs Assessment and Service Review² for the prevalence figures of the main ICCs). The need for services is likely to further increase with the growing knowledge of the molecular basis of conditions and resultant increase in recognition of inherited cardiovascular disease, the identification of individuals who require services arising from cascade testing in families of affected individuals, and the growth in genetic testing as a consequence of advances in technology such as next generation sequencing technologies. The unifying theme for this patient group is the need for a precise genetic diagnosis if possible, along with thorough cardiological investigations, and to provide management together with counselling and advice for patients and families around inheritance issues. This requires both the integration of two specialist disciplines (cardiology and genetics) and the development of shared care between specialist and district cardiology services, with appropriate referrals from primary care.
This Commissioning Guide is produced in the context of the revisions in 2009 of the Specialised Services National Definition Set: 13 Specialised cardiology and cardiac surgery services (adult) (third edition, 2009), 20 Medical genetic services (all ages) (third edition 2009) and 23 Specialist services for children (third edition, 2009) and chapter 8 of the National Service Framework for Coronary Heart Disease (2005). Chapter 8 focuses on quality requirements of services for patients affected by arrhythmias and/or at risk of sudden cardiac death (SCD).

There are 5 main categories of inherited cardiovascular conditions:

1. Arrhythmia syndromes including LQT, short QT and Brugada syndromes and catecholaminergic polymorphic ventricular tachycardia (CPVT)

2. Cardiomyopathies which include hypertrophic cardiomyopathy (HCM) characterised by asymmetrical thickening of the heart muscle thereby obstructing the emptying of blood from the heart and dilated cardiomyopathy (DCM), which weakens the heart muscle. These conditions can lead to heart failure, stroke and arrhythmic defects in heart function

3. Inherited arteriopathies, which cause catastrophic rupture of the blood vessels in addition to affecting other organs. These conditions include Marfan, Ehlers-Danlos and Loeys-Dietz syndromes

4. Muscular dystrophies, a group of multi-system genetic disorders that cause progressive muscle weakness and death of muscle cells. Some muscular dystrophies affect the heart, leading to arrhythmias. Examples include Emery-Dreifuss muscular dystrophy and myotonic dystrophy

5. Familial hypercholesterolaemia, a relatively common monogenic disorder resulting in severe elevation of blood cholesterol and low density lipoprotein, leading to a high risk of premature coronary atherosclerosis which often presents as sudden cardiac death. Being inherited this condition is an inherited cardiovascular condition although it tends to be managed within lipid services rather than cardiology services and so is not considered further in this guide to commissioning of ICC services. Commissioning arrangements for Familial hypercholesterolaemia are also likely to be different in that these services are commissioned by Primary Care Trusts (PCTs) rather than Specialist Commissioning Groups (SCGs). DH are currently working with NHS Primary Care Commissioning to develop a primary care service framework to assist commissioners and others to implement the NICE clinical guideline on identification and management of FH (published in August 2008¹).

A service for patients with inherited cardiovascular conditions will include:

1. Identification of patients with suspected ICCs
2. Appropriate referral which will require clear referral pathways between primary and particularly secondary and tertiary care
3. Diagnosis (including specialist cardiovascular investigation, cardiac pathology and clinical and laboratory genetic elements)
4. Communication and information giving about the condition and its implications
5. Advice and provision of treatment
6. Long term multi-disciplinary support and management
7. A family centred approach
8. Consideration and provision of counselling about risk of recurrence and risk to other family members and
9. Cascade testing which enables family members to be identified as not being at risk and so can be discharged from follow-up or requiring follow-up if they carry the mutation.
Current UK provision

In early 2008, 18 specialist ICC services were identified in the UK along with 2 Marfan services. One of these services, the Heart Hospital in London, sees considerably more patients than any of the other services and receives referrals from across the UK, although the majority of patients are referred from London and the surrounding regions. In the regions, many services are small and operate on a less frequent basis with 2-8 sessions per month. In London, services tend to be larger and have more sessions with services holding 12-44 outpatient sessions per month (12-15.5 if the Heart Hospital sessions are not included). In the Regions only half (7) of the services see more than 200 patients a year while in London all but 1 of the 5 services sees more than 200 patients a year. It is probably the lack of clinical capacity that currently limits the amount of patient activity in smaller services resulting in restricted access to the population as a whole in many SHA areas.

It is unlikely that small services can develop the necessary critical mass to gain sufficient experience of these conditions and to justify the investment in professional training (medical, pathology, genetic counsellor, specialist nurse, laboratory, and cardiac physiology) and organisational development to develop into a fully comprehensive service. Commissioners will need to work together to decide where specialist services will be provided. All Specialist Commissioning Groups should not expect to develop their own service.

Factors that are important in commissioning for service development include:

1. Collaboration of commissioners
2. Involvement of clinical and laboratory service providers
3. Involvement of patient groups
4. The development of a local service specification including local service standards
5. The development of nationally compatible information systems to audit activity against the agreed standards, to audit health outcomes and to monitor access to services by geographic area of residence
6. Timely implementation of evaluated technological developments into service provision.

Current national priorities and initiatives include:

1. The Quality and Productivity challenge
2. World Class Commissioning
3. National Service framework for coronary heart disease and arising from this cardiac networks to develop services and implement the framework locally
4. Delivering the 18 week patient treatment pathway
5. Expert patients programme
6. Transition: getting it right for young people
7. NHS Operating Framework
8. Genetics White Paper
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Service models and modes of organization

Service provision requires both genetics and cardiology input although the relative emphasis may vary according to the expertise available, in particular, in the level of genetics expertise that the cardiologists bring to the clinics and the capacity and supporting cardiac knowledge that can be provided by clinical geneticists. **Joint genetic/cardiology clinics**, with consultant level input in both specialties, is the model currently provided in most centres in the UK and is recognised as being successful. A **cardiology led** service is one where cardiologists with special interest and training in genetics manage most of the patients with the support of a genetic counsellor and/or cardiac genetic nurse and consult with clinical geneticists only regarding those with particularly complex diagnostic, ethical, counselling or interpretive issues. Both models require close working relationships between cardiology and clinical genetics and need to provide family record keeping and family follow-up for effective cascade screening. To address the current under provision of service and anticipated future demands throughout the UK using either model would require a significant increase in capacity, including clinical geneticists and/or genetics-trained cardiologists, genetic counsellors, cardiac genetic nurses and cardiac physiologists. A collaborative approach between cardiology and genetics services would enable the most productive use of resources as endorsed by the QP framework.

The main organizational structures are:

**A hub and spoke model** with referrals from the district spokes to the specialist tertiary hub service. Once patients have been assessed by the hub, much patient care may be devolved to the district spokes according to agreed care guidelines with patients being referred back into the hub where guidelines indicate this is required. Other models include **specialist centres with satellites** in peripheral clinics to enable a greater ease of access by patients who live far from the specialist centre.

Key clinical issues

Key clinical issues in providing an effective service for ICCs include:

**Recognition of possible inherited cardiovascular disease** by general practitioners, adult and paediatric cardiologists and paediatricians with appropriate referral for specialist advice

**Recognition of sudden cardiac death by coroners** (or fiscals in Scotland) with appropriate instructions to pathologists for expert examination

**Expert and timely autopsy diagnosis of index cases** presenting with sudden cardiac death by trained cardiac pathologists

**Ensuring that appropriate referral pathways are in place** and well known and that the specialist inherited cardiovascular services are integrated with other services including primary and secondary care; robust communication channels should be established with those responsible for investigating sudden cardiac deaths - namely coroners, fiscals (in Scotland) and pathologists

**Providing effective and efficient clinical care** of these conditions by **provision of multidisciplinary team** experienced in diagnosis and management of patients and **families** with inherited cardiovascular disorders and ensuring that knowledge of the condition and its implications are cascaded to other family members who may share the same disease causing mutation

**Close involvement of patient organisations** in the planning and delivery of care in order to optimise long term patient care and satisfaction

**Audit and research** to develop the evidence base for effective services and ensure that innovations are translated into clinical practice.
Specifying a service for patients with inherited cardiovascular conditions

Service components include:

1. Ensuring access to specialist services for people with inherited cardiovascular disorders
2. Ensuring timely and accurate diagnosis of people with inherited cardiovascular disorders, including obtaining information from cardiac pathologists who have examined specimens or the bodies of other family members
3. Ensuring access to a high quality prevention, treatment and follow up service
4. Meeting the needs of the family.

Ensuring access to specialist services for people with inherited cardiovascular disorders

Many local clinicians in primary and relevant areas of secondary care currently lack knowledge about inherited cardiovascular disorders and the existence of specialised services for their diagnosis and management.

Commissioners should specify that specialist inherited cardiovascular services should:

1. Work with district services and primary care to develop and implement protocols for referral
2. Devise means of bringing referral protocols and information about services to the attention of relevant professionals
3. Ensure that there are sufficient resources for appropriate genetic testing
4. Promote a family centred service that cascades services to other relevant family members
5. Audit referrals by geographic area of residence and the ethnicity of patient groups
6. Work in conjunction with voluntary organisations to raise awareness of services with patients and families.

Ensuring timely and accurate diagnosis of people with inherited cardiovascular disorders

To make an accurate diagnosis the service should have access to the following:

1. Consultant cardiologist with special interest in inherited conditions
2. Consultant paediatric cardiologist with special interest in inherited conditions
3. Consultant clinical geneticist
4. Genetic counsellor
5. Genetic nurse
6. Laboratory scientist with expertise in molecular genetic testing specialising in cardiovascular conditions
7. Cardiac physiologists including electrophysiologist and echocardiologist with experience of inherited conditions
8. Pathologist with specific interest and training in cardiac pathology and also access to relevant coroner’s records
9. Bereavement and counselling services for those who have suffered a bereavement.

These individuals should function as a multi-disciplinary team with joint input into diagnosis.
Investigations, such as the following, should be available to support the diagnostic process:

1. Specialised electrophysiology including signal averaged ECG and exercise ECG, drug provocation testing, holter monitoring, echocardiography, cardiac MRI, cardiopulmonary exercise test and tilt testing
2. Ophthalmology testing for Marfan syndrome
3. Molecular genetic testing, including conditions relevant to cardiovascular disorders listed by the UKGTN.

Genetic testing aims to identify the particular mutation in the proband (first individual to contact services) with testing of further family members for that particular mutation only. As a result, other family members can either be identified as not being at risk and discharged or identified as requiring further monitoring. The identification of a mutation is usually necessary if prenatal testing is to be undertaken.

Commissioners should require services to specify:

1. How they will ensure services are aware of and can access UK Genetic Testing Network (UKGTN) listed tests
2. The adoption of UKGTN testing criteria or other approved referral criteria for genetic testing
3. How services will decide who can request a genetic test and relevant education or other support for those individuals
4. The methods and personnel involved in gate-keeping
5. How commissioners and services will monitor equity of access to testing.

**Ensuring access to a high quality prevention, treatment and follow up service for patients**

Once diagnosis of an inherited cardiovascular disorder is made patients are likely to require long term follow up. Although some routine aspects of care might take place nearer to their homes, in general, patients value long term management by a consultant with expertise in the particular condition who they trust will have pertinent information about the best methods of treatment and keep up to date with advances in clinical management. They also need to have continuing contact with services as their own circumstances evolve - for example, other family members, including their own children, might need testing or reproductive advice.

The specific preventive, treatment and follow up services are particular to the disease in question and are many and varied. Examples of particular interventions include:

1. Alcohol septal ablation or surgical myectomy in a subset of patients with HCM who have obstructive thickening of a part of the heart muscle
2. Genetic testing to identify sub-types of LQTS to enable appropriate drug therapy to be instigated; for example certain types of LQT (LQT1) respond well to beta blockers while other types (LQT3) do not respond well to this type of drug
3. Preventive advice tailored to the patient as a result of the refinement in diagnosis provided by genetic testing. For example LQT1 is more frequently triggered by exercise and so certain types of exercise should be avoided while LQT2 is more commonly provoked when the patient is roused by a sudden noise while sleeping or at rest so use of alarm clocks and the like should be avoided
4. Initiation of statin therapy which is effective for patients with FH.
Commissioners should require services to specify:

1. How wider multi-disciplinary input into management plans is provided and coordinated
2. How referral and shared care protocols are operating
3. How patients are enabled to contribute to and take responsibility for aspects of their own management plans
4. How patients are followed up by the specialist service long-term
5. How arrangements are made for routine care closer to home
6. How care is managed as a ‘one-stop’ shop so that patients need not make repeated visits for different aspects of diagnosis or management
7. How transitional arrangements between paediatric and adult care are functioning
8. How patients and families are made aware of support provided by voluntary sector.

Meeting the needs of the family

The family needs an inherited cardiovascular service that identifies and follows up family members who might also be affected or who might be at risk of having an affected child.

The ability to link patients and individuals in families through family records is also useful in achieving the most efficient means of testing where multiple family members have contacted different services at different times from different geographic locations.

Many services are currently run by joint genetic/cardiology services and the family record keeping is undertaken within the genetics department. At present this is a regional rather than a national service.

Commissioners should ensure that services set out:

1. How family records will be kept and which service is responsible (genetics or cardiology). This should include necessary elements of consent and confidentiality and what links will be made between the genetic and cardiology records
2. How the decision on family cascade testing is initiated and undertaken and who takes responsibility for it. This should include how relatives are approached and how any complex ethical issues in contacting relatives (such as estranged or adopted relatives) are resolved
3. How services are alerted to testing that might have taken place for another family member.
Commissioning a service for the management of inherited cardiovascular conditions

Determining Local Service Levels

Benchmarking data are based on epidemiological data and information derived from a survey of UK inherited cardiovascular services².

Information on estimated regional service requirements can be based on London and SE Coast regional figures. Services in London and SE Coast see 204.23 new patients per million population. From this figure an estimate of local service need can be calculated based on the regional population. For a standard primary care trust population of around 250,000, or an average practice (list size 10,000), it is thus clear that such activity would not justify provision of a specialised service. For an average Strategic Health Authority, with a population of around 5 million the expected level of service requirement would amount to about 1,000 new patients per year as well as further service provision for the follow-up of patients that require this to be undertaken by the specialist service rather than the district service.

For the UK as a whole it is estimated that about 12,500 new patients would be seen each year. This represents a current shortfall of around 7,000 patients as approximately 5,774 new patients were seen by services as estimated by specialist services across the UK². This is likely to still be an underestimate of need as suggested by the epidemiological data presented in Heart to Heart Inherited Cardiovascular Conditions Services: A Needs Assessment and Service Review². There is no epidemiological evidence of ‘incidence’ of ICCs in terms of the numbers who would be likely to present clinically each year, nor do we have evidence on the overall number of those presenting who would need regular surveillance and at what level.

Based on the London services activity rates from a survey² of the UK inherited cardiovascular services (using figures from all London services), a service seeing 1,000 new patients per year would need to provide 386 outpatient sessions per year or 32 sessions per month. Again using London figures, this would require 825 consultant sessions per year or 69 total consultant sessions per month.
Figures for our bench marking were derived from the service survey².

Estimated requirements for provision for 1000 patients (the estimated rate for an SHA of 5 million) was derived from data from our survey² on average annual outpatient clinic sessions and consultant sessions as delivered at the London services (Northwick Park, London Chest Hospital, Guys and St Thomas’, Kings and Lewisham, St George’s and The Heart Hospitals - these services serve 3,088 new patients per year).

<table>
<thead>
<tr>
<th>Services</th>
<th>Estimated annual number of new patients seen</th>
<th>Annual outpatient sessions*</th>
<th>Average annual consultant sessions*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Northwick Park</td>
<td>700</td>
<td>186</td>
<td>234</td>
</tr>
<tr>
<td>London Chest Hospital</td>
<td>130</td>
<td>168</td>
<td>168</td>
</tr>
<tr>
<td>Guys and St Thomas’, Kings and Lewisham</td>
<td>630</td>
<td>168</td>
<td>240</td>
</tr>
<tr>
<td>St George’s</td>
<td>419</td>
<td>144</td>
<td>288</td>
</tr>
<tr>
<td>The Heart Hospital</td>
<td>1209</td>
<td>528</td>
<td>1620</td>
</tr>
<tr>
<td>Total</td>
<td>3088</td>
<td>1194</td>
<td>2550</td>
</tr>
</tbody>
</table>

| Annual number of OP sessions per 1000 patients   | Monthly number of OP sessions per 1000 patients | 32                          |
| Annual number of consultant sessions per 1000 patients | Monthly number of consultant sessions per 1,000 patients | 69                          |

* 1 session equals one half day

There are published guidelines³ on when genetic testing should be undertaken and which tests should be performed. Further data on the number of genetic tests that would be required in the diagnosis of this number of patients, or the level of other specialist investigations would be helpful. The survey undertaken by the PHG Foundation² suggests there are currently very variable levels of genetic testing with more than a 20 fold variation between regions with the most and least genetic testing per million population. This translated to ranges of 1 in 1 patients having a genetic test to 1 in 24 patients. With the rapid advances in genetic testing technologies, the demand for genetic testing and the clinical utility conferred, the demand for genetic testing is likely to markedly increase.
For all these figures, it should be noted that this is likely to be a minimum level of service requirement. Services noted upward trends in patient referrals and, it is likely that this trend will continue because of increasing awareness, further development of diagnostic and treatment options and systematic identification of family members also at risk.

Other Initiatives

**Payment by Results (PbR) project on ICC services**

To ensure that the payment structure for ICC services better reflects their complexity and the range of professionals required to deliver a high quality service, a payment by results (PbR) project is being undertaken at St George’s Hospital, London, in partnership with the SW London cardiac and stroke network, to develop a tariff for ICC services. The project is developing a tariff model based on the existing cardiac outpatient tariff taking into account the need for a multi-disciplinary approach and additional diagnostics specific to ICC patients such as genetic testing.

The tariff model being developed at St George’s Hospital includes pre-assessment by the cardiac genetic nurse followed by an appointment in the clinic to see at least one member of the multi-disciplinary team: consultant cardiologist; consultant paediatric cardiologist; consultant medical geneticist; and/or genetic counsellor. Investigations undertaken routinely include an ECG, signal averaged ECG, echocardiogram, exercise test and 24 hour tape. Additional tests are then considered for diagnostic and risk stratification requirements including an ajmaline test, a cardiac MRI scan and genetic tests. Since the needs of patients will differ according to their clinical and non-clinical circumstances, different packages of care are being defined and costed. Once the tariff packages have been defined, they will be examined to see whether they are sufficiently robust and whether they are suitable for becoming a national tariff or whether they are only applicable as a local tariff. The report on this work is expected in early 2010.

**BHF Genetic Information Service**

The BHF Genetic Information Service (GIS) helps families affected by an inherited heart condition to access clinics for inherited heart conditions. A letter is sent to the family GP supporting the need for referral with the contact details of the clinic(s) in the local area. The GIS is staffed by cardiac nurses who can also provide information on inherited heart conditions and details of other charities who can provide support. The service is open from 9am to 5pm Monday to Friday and is available by calling 0300 456 8383. A range of booklets on inherited heart conditions are available free of charge.
World Class Commissioning competences

Commissioners should work towards the following World Class Commissioning competences to provide key benefits for patients:

**Competency 2:** Work with community partners by:

- Ensuring good communication and liaison with other support services including social services, disability support services, employment services, housing, careers advice and the voluntary sector.

To provide:

**Enhanced long term management** of patients and families with good patient follow-up and robust links with relevant health and other services.

**Competency 3:** Engage with public and patients by:

- Involving patients in service planning
- Developing effective methods for two-way communication between professionals and patients
- Developing close working arrangements with supporting voluntary organisations to provide support for patients and families.

To provide:

**Improved overall quality of life** for patient and family by the provision of accurate and trusted information on prognosis in order to support life decisions that maintain social and psychological well being.

**Competency 4:** Collaborate with clinicians by:

- Commissioning multidisciplinary teams including geneticist, adult and paediatric cardiologists, genetic counsellor, genetic nurse, laboratory scientist, cardiac physiologists and specialist cardiac pathologist
- Developing services which are integrated with other components of patient care including other specialist services (particularly important where the disease is multisystem) and with primary care and secondary elements of cardiology services
- Providing training for staff to achieve relevant competencies
- Developing effective referral protocols and shared care arrangements.

To provide:

**Increased patient satisfaction and trust** that they will be managed by a team with experience in their particular condition

**Improved clinical outcomes** by provision of the most effective preventive and treatment options to slow disease progression and reduce the risk of SCD (through knowledge and appropriate liaison with other specialties) in other organ systems involved

**More efficient and effective care** by the formalised coordination of expertise from the two main specialities involved (cardiology and genetics)

**Enhancing reproductive choice** for parents by providing information on recurrence risk of conditions for future children and providing counselling on reproductive options.
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Competency 5: Manage knowledge and assess needs by:
- Recognising the need to balance specialisation with reasonable geographic accessibility
- Audit and research of service activity.

To provide:
- Reduced inequalities by the development of services that are accessible to all geographic areas and outreach, through general cardiology services, to all patient groups.

Competency 6: Prioritise investment by:
- Developing systems of decision making for the use, interpretation and prioritisation of genetic testing.

To ensure:
- Efficient and effective use of genetic testing.

Competency 8: Promote improvement and innovation by:
- Provision of professional education and training including awareness-raising in primary and secondary care.

To provide:
- Efficient referral of patients to the most appropriate specialty in a timely manner.

Competency 10: Manage the local health system by:
- Ensuring good communication and transitional arrangements between paediatric and adult care
- Developing systems for long term follow up including systems for initiating and undertaking family cascade testing and family record-keeping.

To provide:
- Continuity of care for all family members at risk or affected by an inherited cardiovascular condition.
Commissioning Guide: *Services for patients with inherited cardiovascular conditions*

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**Further Information**

