Do Once and Share

Clinical Genetics Project Report

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Do Once and Share: Clinical Genetics Project Report

1.0 Executive summary

The Do Once and Share (DOAS) programme is part of NHS Connecting for Health. The programme aims to enhance and inform the development of the new NHS IT systems to meet the up-to-date requirements of health services for patients. This report details current clinical practice, projected requirements and recommendations for the NHS Clinical Genetics Service in England following extensive professional and patient consultation.

The work documents a nationally agreed care pathway and accompanying dataset that records the patient journey through the clinical genetics service identifying key IT needs. In addition, the report highlights potential ethical constraints with the use of electronic records in genetics practice and knowledge requirements of patients and staff.

1.1 Benefits of a nationally agreed care pathway and dataset

The benefits include:

- Clarification of an agreed 'patient route' through the clinical genetics service from referral to discharge for the benefit of patients and health professionals using the service
- Ability to collect comparative audit data to demonstrate equity of access and standards of care
- Possibility of a collective national minimal clinical dataset for clinical research and education
- Easy dissemination of new models of service development between centres
- Standard 'unit of currency' for commissioning services
- Clear demonstration of the 'points of contact' with other specialties
- Identification of required access to knowledge support for professionals and patients

1.2 The NHS Clinical Genetics Service

Clinical genetics services care for patients, relatives and health professionals seeking advice about disorders with a genetic, or potentially genetic, cause. Such conditions affect all age groups from the fetus to the elderly; can involve all body systems and range from human developmental abnormalities to cancer. In the last 20 years, the ability to use genetic information for the benefit of patients has changed significantly. Health professionals working in the specialty use the advances made in identifying single gene aberrations, mutations and copy number changes, to make an impact on the health of individuals through diagnosis and classification of disease to inform patient management. This is genetic medicine. While there is a tendency to assume that genetic medicine deals with rare multi - system conditions such as, Neurofibromatosis, a rare genetic disorder of the skin and nervous system, there are Mendelian subsets within common disorders. For example, BRCA1 and BRCA2 subgroups within breast cancer make up a significant part of the clinical genetics workload.

Genetic medicine is well established. In England, it is delivered by the NHS through 18 specialist Regional Genetics Centres and has brought important benefits to the families seeking care. Each centre employs a multidisciplinary team involving doctors, counsellors, administrative staff and scientists and covers a population of between 3 and 5 million people. The clinical component is an outpatient-based speciality although patients are also seen on the wards, in other locations including GP surgeries, and at the family home. Most genetic testing in the NHS is performed within the regional genetic laboratories.

Clinical genetics services recognise that individuals are part of families and genes have a predictable inheritance pattern. In clinical genetics, the fundamental unit of responsibility is the ‘family’ and includes not only the ‘sick’ individual who presents for diagnosis and treatment but also the family members who are identified as being at-risk, i.e. while an individual who presents with ill health needs to be diagnosed and treated within the traditional NHS model, the awareness of family and the relationships within it mean, that the ‘at- risk-but-well’ relatives can be managed appropriately offering the opportunity of prevention and early intervention. This is an example of targeted prevention.

1.2.1 Role of the Clinical Genetics Service

The clinical genetics service may be asked to play a part in:

- Diagnosis (including prenatal and pre-symptomatic diagnosis)
- Prognosis and natural history of genetic disorders
- Appropriate clinical management
- Genetic risk assessment
- Family working to identify the 'at-risk-but-well'
- A 'genetic pathology role' – identification and interpretation of genetic tests
- Translation of research findings into clinical practice and initiating research from clinical observation
- Education of patients, families and health professionals

1.2.2 Tools used by the Clinical Genetics Service

Tools used include:

- Family history taking
- Painstaking information gathering
- Medical history and examination
- Clinical investigation
- Genetic testing

1.2.3 Examples of referrals to the Clinical Genetics Service

A child is noted to have delayed development

Clinical genetics services may play a part in diagnosis, genetic testing, advice on prognosis surveillance, treatment strategies, risks for relatives, risks for unborn children and prenatal diagnosis.

A mother develops breast cancer at 40 years

Clinical genetics services may be asked a) the likelihood that the cancer is due to a 'genetic subset' of breast cancer, b) the risks that her children may develop breast cancer, c) appropriate screening for her children and d) the best available breast cancer management for the patient.

A patient is diagnosed with a dilated aorta and dies of a ruptured aneurysm

Clinical genetics services may be asked if there are genetic investigations to identify what was the cause and who else is at-risk in the family.

1.2.4 Integrating genetics into medicine

While clinical genetics may be considered a part of 'medicine' the reality is that medicine is actually a part of genetics and the clinical genetics Do Once and Share team have established the care pathway for genetic medicine. The pathways and skill mix identified in clinical genetics will have relevance to all specialities where Mendelian subsets of common disorders can be shown to exist.

1.3 Key findings of the Do Once and Share Clinical Genetics Project

1.3.1 Unit of responsibility in Clinical Genetics is the family

- Individuals are part of families. A family encompasses both a genetic and a social relationship.
- The unit of responsibility in clinical genetics is the 'family', which leads to, targeted prevention of the at-risk family members.
- Geneticists have a legitimate relationship with patients not under their direct care by way of their relationship with another relative
- Not all patients that are seen in clinical genetics services are referred. This results in 'Episodes of Care' for relevant family members of the index case identified to be at-risk, a form of targeted prevention.
- Family notes are used to record information that has relevance to more than one family member of the family.

- There is universal use of 'family notes' within UK Clinical Genetics Services. They avoid repetition, are efficient and prevent false diagnosis where information from one family member is relevant to another.
- Family history is a tool used universally in all clinical genetics practice resulting in information from other relatives being included in the records.
- Family information and personal pedigrees are used by all specialties practicing genetic medicine, which means that generic tools are needed for pedigree drawing.
- A distinction is made between 'personal pedigree'- defined as the knowledge a person has of their own family history and a 'modified genetics pedigree' – defined as family history where the findings have been verified by the genetics service.

1.3.2 Core of the Clinical Genetics Care Pathway: the 'Clinical Genetics Episode of Care'

  - An outpatient appointment (or Significant Contact replacing an outpatient appointment)

  - An Episode of Interaction/Advice – a non-planned piece of Interaction/Advice not equivalent to an outpatient appointment

  - A Finished Clinical Genetics Episode of Care (FCGE). This broadly equates to the care required to answer the patient referral reason and may be made up of more than one outpatient appointment (or Significant Contact) and sometimes also involves an 'Episode(s) of Interaction/Advice

- There are 3 major clinical components to a Finished Episode of Care which is often a synthesis of all three; information gathering, medical consultation and genetic counselling. These may all be covered in one outpatient appointment/'Significant Contact' or several appointments may be necessary before a definitive clinical genetics opinion can be given. The episode is not termed 'finished' until the patient question is answered or a professional decision is made that nothing further can be offered
- The 'Finished Clinical Genetics Episode of Care' (FCGE) can be used as a clinical genetics unit of currency if necessary and provides a more realistic measure of the complexity of the care provided in clinical genetics than using an 'outpatient appointment' as the unit

- Care episodes can be offered in clinic, on the ward, at the home of a patient, on the telephone or electronically and take differing lengths of time. They are all legitimate with different costings but the decision is a professional one. Some components of care can only occur in a face-to-face situation e.g. clinical examination
- There are currently three categories of health workers in clinical genetics: doctors called clinical geneticists, genetic counsellors and administrative staff. All have different skills and different training. They work synergistically for the benefit of patients
- Follow-up and ongoing management of families is divided into follow-up 'Episodes of Care' and /or 'Episodes of Interaction /Advice'. Coding provides the possibility of distinguishing new from follow-up 'Episodes'
- 'Episodes of Care' may be provided for relevant relatives of the referred individual if they receive significant personal genetic advice usually warranting separate or additional communication to another health professional
- The dataset is designed to reflect the granularity of the service and capture information relevant for research, audit and costings

1.3.3 Clinical Genetics interface with other specialties

- Investigations are requested for the full range of pathology tests, radiology, and particularly genetic testing from the NHS and specialist oversees genetic laboratories
- Referral to other specialties for specific investigations or clinical opinion such as echocardiography and eye examination
- Investigations in clinical genetics services include gathering relevant family information, pathology reports of relatives, recorded notes including death certificates and are termed additional information in the care pathway and dataset
- Touch points with other specialties include:

    - Open-for-access discussion with all specialties in primary, secondary and tertiary care, patients and family members

    - Referral to other specialists for specific investigations/specific system examination

    - Referral onwards to other specialties for continuing medical care

- Joint working with other specialities in combined clinics where the care pathway will function alongside other specialty care pathways

- Identification of 'points' within other specialty care pathways where they should connect with the Clinical Genetics Care Pathway through agreed referral guidelines, management guidelines and genetic testing requests

1.4 Recommendations

1.4.1 Electronic family notes and pedigree drawing

Consultation for this project provided strong support for the need for family notes to continue in Clinical genetics services and recommended:

- Family notes should be 'housed' within clinical genetics services and contain the 'modified genetic pedigree' and information on more than one relative where appropriate
- Family notes to exist alongside the NHS personal electronic notes
- Include in the NHS personal electronic notes, the family notes number that will denote both the genetic family number and the genetics centre holding the notes e.g. SOT123444 for a Southampton family record
- A legitimate relationship between trained genetics health professionals and relatives of index cases
- An electronic system that could allow access, by trained genetics health professionals, to medical records of relatives with robust and appropriate consent
- There are two proposed models that need to be debated openly with the Care Record Development Board, by public debate and with the Human Genetics Commission:

    - ***'Family Opt Out'*** – NHS patients allow all relevant health information to be accessed by genetic health professionals caring for relatives, with the option to 'hide' personal data they are not willing to share

    - ***'Family Opt In'*** - Patients to transfer specific health data to a 'genetics/family envelope' for sharing with relatives

- Electronic family history pedigree drawing software should be available for all health The dataset for the personal pedigree information needs to be agreed and is not part of this report
- Electronic family notes are required if the clinical genetics service is to be 'paperless'

1.4.2 Electronic data collection and knowledge transfer

- There are no universal diagnostic or 'issue' codes for genetic disorders in the UK SNOMED-CT/READ codes have not yet included many of the rare genetic condition codes. ICD codes do not meet the diagnostic needs of clinical genetics. Coding for genetic disorders needs to be addressed in England
- A national collection of data is an opportunity to bring together information collected from genetics clinics in the UK. Given that many of the disorders encountered are rare genetic conditions, such a unique collection of data could provide information on natural history, identify key medical complications and be a powerful clinical research tool. At present such data are collected individually in paper records but cannot be pooled and the collective value is 'lost'
- Health professionals dealing with rare conditions are particularly in need of up-to-date electronic access to current knowledge
- It is recognised that clinical genetics services require on line access to many contemporary peer reviewed genetics journals that are not currently available to the NHS on line. It is thus envisaged that the diagnostic code will lead to automatic IT links to:

    - A minimal clinical dataset for the condition requesting entry of specific data e.g. aortic dilatation by age in Marfan syndrome, or head circumference by age in PTEN hamartoma syndrome. In time this may be collected automatically from electronic entry into the notes of particular features

    - Clinical management guidelines for diagnosis on on-going management. N.B. Many diagnostic and management guidelines are not yet written for these types of disorders as data are lacking, but are required

    - Support group literature

    - Information leaflets for patients, including the need for translated information, where appropriate

- The most recent reviews and peer reviewed papers. Note the NHS access to genetic journals is inadequate for clinical practice at present

- UKGTN (UK Genetics Testing Network) genetic testing availability

- The gene structure, mutation database, DECIPHER, tissue expression pattern, and disease associated mouse models

- On going research studies such that patients can decide if they wish to participate

- Genetics databases such as OMIM, DECIPHER and London Dysmorphology database

Interface with other specialties

- Identify where the clinical genetics pathway interfaces with other speciality care pathways when they written
- Write referral guidelines to Clinical Genetics Services

Dataset for Clinical Genetics Services

- Codes are required that link the sequence of appointments within an Episode of Care, the sequence of Episodes of Care and the sequence of Episodes of Interaction/Advice either within an Episode of Care or at other times independent of it, with the patient number and family number. Attempts have been made within the dataset to suggest coding systems but these need to be reviewed by IT code experts
- There is a need for a unit of currency in clinical genetics for audit and commissioning purposes but this is not simple. Counting outpatient appointment numbers is one method but one outpatient appointment is not necessarily comparable with another and by trying to provide a simple measure of the service its complexity may be lost. For example some are information-gathering appointments and others are medical consultations. Both may be required to deal with the patient query but neither would be sufficient alone. DOAS Clinical genetics recommends that the 'unit of care' is the 'Finished Clinical Genetics Episode of Care' (FCGE) as this attempts to convey the complexity of the care given in clinical genetics. Given that each 'contact' is recorded in detail within the dataset, the

make up of the FCGE can be easily identified if that is necessary for audit and cost purposes. A comparison of this approach with that used in other specialties would be useful

2.0 Main Components of the Clinical Genetics Care Pathway

2.1 Clinical Genetics Episode of Care

The Clinical Genetics Episode of Care is made up of one, or a series, of outpatient appointment(s) or equivalent, termed Significant Contact(s) and has 3 broad clinical components designed to answer the referral reason: information gathering, genetic counselling and medical consultation. Any aspect of these may be necessary in addition to investigations, additional information gathering and referral for further specialist opinion to complete an Episode of Care

An outpatient appointment is defined as a planned face-to-face meeting between a patient and genetics health professional in hospital. At times it may be replaced by other types of 'Significant Contact' including:

- Face to face contacts at home, on the ward
- Letters, phone contacts, electronic contacts that replace a face-to-face clinic contact

  But excluding, for the purposes of this definition:
- Contacts not replacing an outpatient appointment

An 'Episode of Care' usually begins with a referral of an individual. A geneticist (counsellor or doctor) contacts the family and arranges an outpatient appointment or uses alternative methods such as a telephone consultation to gather information. The history taking will include information about the presenting issue, as well as a detailed family history. Permission is often required from family members to verify diagnosis including pathology results and genetic testing. The tracing of death certificates may also be necessary. This verification process takes time. It may be possible to combine the components of the consultation in one appointment (if all the information is available) but it may mean that a second appointment is required. Sometimes more than two outpatient episodes will be necessary before the referral reason is answered particularly, where the episode requires investigation of the consultand such as genetic testing.

2.2 Finished Clinical Genetics Episode of Care (FCGE)

The currency of care is termed a 'Finished Clinical Genetics Episode of Care'. More than one outpatient appointment or Significant Contact with different members of the genetics team may be necessary before it is termed 'Finished'. The decision that the Episode of Care is 'Finished' is a professional one.

2.2.1 Family case scenarios

Example 1: A woman (consultand) has a family history of Huntington's Disease and requests a pre-symptomatic genetic test.

Following triage of the referral, the patient is sent an appointment to be seen in clinic, Significant Contact '1'. The family history is taken. Attempts to verify the diagnosis in at least one affected family member is initiated by arranging with the consultand to contact the affected family member for consent to access the relevant records. This is important as there are other disorders that can resemble Huntington's Disease, and a genetic test for this condition will only diagnose/exclude this specific condition. A family history will be used to calculate the risk for the consultand of inheriting the disease. It is important to discuss the natural history of the condition, the reliability of the test and the implications of the result. Time, for consent and verification of diagnosis in the family member, is required, as well as the opportunity for the patient to reconsider the desire to be tested. There will be communication after this contact to the patient and the referring health professional. In some situations the Episode of Care will 'finish' after this initial episode if, following the consultation, the patient decides to take no further action.

In other instances, the patient will be keen to progress to genetic testing. This will be undertaken at Significant Contact 2. A date will be planned taking into consideration the expected date when the confirmation of diagnosis is likely to be available. It must be noted that delay is often outside the control of the genetics department. This second contact may include a neurological examination and further genetic counselling to ensure that the patient will not regret the test and has fully appreciated the ramifications of pre-symptomatic testing. Blood extraction is performed at Significant Contact 3, and the sample sent to the genetics laboratory for testing for Huntington's Disease. A date will be arranged for result giving at Significant Contact 4. Communication will be sent confirming the result to the patient and referrer. Thus four Significant Contacts (equivalent to 4 outpatient appointments) are involved in this 'Finished Clinical Genetics Episode of Care' with different members of the genetics team.

Sometimes the consequences of information giving need further immediate significant 'contacts'. It will be a professional decision whether to include these as part of the first 'Finished Episode of Care' or as a separate follow-up 'Episode of Care'. For example, a follow up within 24 hours because of the impact of the test result, may be included within the first 'Episode of Care'. A planned follow-up in 4 months to assess any neurological progression of the disease would be

considered a review and thus a second 'Episode of Care'. In practice, funding decisions may dictate which of these are used. The dataset allows time taken, by whom, with whom and where it is undertaken, to be recorded, irrespective of whether it is part of Finished Episode of Care 1 or 2. A team approach is used to maximise efficiency. Experienced genetic counsellors perform information gathering and genetic counselling; a genetic doctor will undertake the medical examination and discuss the detailed prognosis. Appropriate planning allows for appropriate case mix.

The family nature of this type of care can be easily appreciated. A positive Huntington's Disease result in this example may lead to other family members requiring a genetic referral. For example, an adult son of a woman testing positive for Huntington's Disease, faces a 50% risk of inheriting the gene and may seek referral himself. It must be noted that the information from this woman will be of major significance to her son's risk and her genetic test result will provide confirmation of diagnosis for the son. Hence, the results from the woman, with her permission, are recorded in 'family notes' that will be used subsequently for all family members. In other instances, the woman may have young children who will require genetic testing in the future. (Huntington's Disease testing is usually offered to adults over the age of 18 years). Her health information is therefore of lifelong significance for her future descendents, requiring lifelong access and availability.

She may have relatives at-risk living in other areas of the country and therefore, with her permission, her test results may be needed for verification of diagnosis by other genetics centres. The initial referral of the woman may lead to the Genetics Service offering 'Episodes of Care' to other family members not originally referred.

Example 2: A woman (consultand) is referred with a family history of breast cancer

Significant Contact 1 may be undertaken by telephone between a genetic counsellor and the patient. It involves history gathering, diagnosis verification and subsequent correspondence to family members for consent to access patient notes. Cause of death of relatives from death certificates will be sought. In this example, genetic testing will be initiated in an affected family member before testing is possible for the consultand. This illustrates the importance of the relationship between the geneticist and the family rather than just the referred patient. There is a legitimate relationship between the clinical genetic health professionals and relatives of an index case and this is part of normal NHS Clinical Genetics practice.

Synthesis of the information gathered by the genetics team will result in a second 'Significant Contact' either involving medical history, examination and potential genetic testing of the

consultand, if the genetic breast cancer risk is high, or if low, the information giving may be provided in alternative settings by a telephone call or by letter. Different team members can deal with different aspects of this 'Episode of Care'. Significant 'contact' '3' may be required for genetic test result giving before the episode is considered 'finished'.

2.2.2 Clinical Genetics DOAS recommendations for variability in interpretation

There will be differences in the interpretation of how to count Significant Contacts within the 'Episodes of Care'. For example in example one, where a patient has 4 Significant Contacts to deal with the initial enquiry for testing for Huntington's Disease, some may count this as 4 Episodes of Care and others as one. The outcome of the Do Once and Share Clinical Genetics project recommends that where possible a 'Finished Clinical Genetics Episode of Care' (FCGE) is the 'unit of care' used to try and answer the 'patient referral question'. For example, a patient requests a 'genetic test for Huntington's Disease' and it takes 3 or 4 Significant Contacts to deal with the question. A time lapse of more than 1 year between Significant Contacts will be considered a new Episode of Care but this decision can be reviewed as necessary. In another example if a patient requests a genetic risk assessment for schizophrenia, it may be clear that only one Significant Contact is required within the Finished Clinical Genetics Episode (FCGE) to answer the question. Given that all details pertaining to the consultation are recorded in the dataset, this will allow easy analysis of the way the FCGE is being interpreted by each centre for the purposes of payment and audit.

The Episode of Care may involve genetic testing. Some of this is relatively routine and takes place in an NHS Genetics laboratory. The time taken to finish the Episode of Care may include the time taken for the test to be performed and reported. However there are occasions where samples are sent to research laboratories and the time taken for the blood to be tested can be very long (up to years). It will be a professional judgement whether this will be dealt with under the 'review' section of the pathway or within an 'Episode of Care'.

2.3 Episode of Interaction/Advice

Clinical genetics services, in most regions, are set up so that patients and/or health professionals may seek advice directly from the service usually by telephone. If the enquiry can be dealt with entirely within the 'two-way episode of communication' this is termed an 'Episode of Interaction/Advice'. The discussion needs to be logged so that the information discussed can be found, should the patient/health professional re-contact the department. The dataset that

accompanies the episode, records key data allowing searching of patient name, dates and issues discussed. There will be occasions when the advice will not be patient related so the dataset includes the ability to not include patient identifiers if not appropriate. This method of information giving is particularly pertinent for a speciality where a key component of health benefit is information and this type of advice forms a significant part of the workload of a clinical genetics service. It needs to be recorded in addition to patient activity in the outpatients.

2.3.1 Family case scenarios

Example 1: A General Practitioner phones the Clinical Genetics department because he has seen a pregnant woman who has a sister with Down's syndrome. His patient wants to know the risks that her unborn child will have Down's syndrome?

The GP is able to tell the geneticist that the sister with Down's syndrome has Trisomy 21 (three copies of chromosome 21, instead of the normal 2). The geneticist can inform the GP that the risks for the woman are not increased as a result of the family history and are equivalent to any woman of her age. The concern is therefore dealt with, without the need for a formal referral or an outpatient appointment. However, details on the advice given are recorded in case of further requests by the family in the future, and for legal and audit reasons.

Example 2: A man phones the Clinical Genetics department because he has just learnt that his nephew, who has severe muscle weakness, has been diagnosed with Duchenne Muscular Dystrophy. He is aware that this is a genetic condition and wants to know if his own son will get the disease?

Provided the diagnosis is correct, the risk to the man's child is low because the condition is due to a gene fault (gene mutation) on the X chromosome. Males with the faulty gene are affected; females with one copy of the faulty gene are usually asymptomatic carriers. Therefore, if the man in question is healthy and has no muscle weakness, he could not have a copy of the 'faulty' gene. In addition as males do not pass their X chromosome to their sons, the risks to his son are low. The rapid giving of this information may provide reassurance to the patient and is a more efficient way of providing advice than offering an outpatient appointment. This type of information giving is increasingly popular as communication between health professionals and patients improves. This needs to be documented over and above outpatient activity, hence the inclusion of an Episode of Interaction /Advice in the Care Pathway. This enquiry from one family member may initiate referral of other members of the family and the work involving this initial enquiry needs to be recorded so

that it can be used in subsequent encounters with the family. It should be noted that the information given in example 2 is only relevant if the disease has been correctly diagnosed and the man is indeed healthy with no evidence of muscle weakness. In the future this information may be immediately verified if the electronic records of the man in question are available to the geneticist. Likewise the definitive diagnosis and molecular results on his nephew may also be available if the nephew and his family have consented that these are accessible to family members.

Alternatively, if there remains some uncertainty about the diagnosis it may be viewed appropriate to verify the diagnosis in the nephew and examine the man. This will involve thorough family discussion, information gathering, medical consultation and genetic counselling. The initial enquiry might therefore lead to a referral to the clinical genetics service. Usually the patient will be asked to contact their GP and request referral but clinical genetics services will accept self-referrals if there are legitimate reasons why the patient does not want to inform the GP. (In future, the commissioning process will govern this and determine whether funding for self-referrals is available).

2.3.2 Clinical Genetics DOAS recommendations for variation in interpretation

It may be difficult to be certain whether an Episode of Interaction/Advice constitutes equivalence to an outpatient appointment or not. This unit of care is to be used for ad hoc and emergency requests for advice. Planned episodes of two way communication and face-to-face contacts constitute an Episode of Care rather than an Episode of Interaction.

During the project consultation period, there was confusion about how to record a referral that involved work to decide whether a referral was necessary. For example, a referral for breast cancer risk assessment, that involves prior information gathering to show whether the genetic breast cancer risk to the consultand is low or not. The Do Once and Share Clinical Genetics recommend that this clinical activity, which will include advice to the patient and referrer, is considered equivalent to an outpatient appointment and is therefore an Episode of Care. It is not deemed an 'Episode of Interaction' as it is not an ad hoc communication, rather a summation of information and a response to a referral.

2.4 The unit of responsibility in Clinical Genetics is the 'family'

When a patient is referred to the genetics service the patient referral may lead to an Episode of Care for another member(s) of the family, in addition to the referred patient. This is because genetic conditions are inherited through the family in predictable patterns and the diagnosis of a genetic disorder in one family member means that others may be predicted to be at-risk. While there are parallels with other specialities, for example paediatrics, where a child is referred and the parents are counselled on the consequences of the condition, in genetic medicine the consultation often has direct relevance to the health of the family member.

2.4.1 Family case scenarios

> ***Example 1: A teenager is referred with a possible diagnosis of Marfan syndrome (an autosomal dominantly inherited genetic condition predisposing to aortic aneurysm) for confirmation and discussion of appropriate medical management.***

His mother accompanies him in the clinic. A family history reveals that his grandfather, the mother's father, died at the age of 37 years of a ruptured aneurysm. It is possible, therefore, that the grandfather too had Marfan syndrome. A diagnosis of Marfan syndrome in the son makes it likely that the mother must have inherited the gene for Marfan syndrome from her father and passed it on to her son. This makes the mother also at-risk of aortic aneurysm. Therefore, one referral generates two 'Episodes of Care' on both the son and the mother and referral on to the cardiologists of two people. Dependent on the age, they may be referred to two different cardiac specialists.

In addition to this example, where the relevant family member is seen because they are found to be at-risk, there are other occasions when relevant family members will be asked to attend because the examination of relatives can be of importance in the diagnosis of the index case.

> ***Example 2: A child is referred with possible Neurofibromatosis (a genetic condition of the skin and nervous system, predisposing to the formation of tumours) for diagnosis and advice on medical management.***

The diagnosis of Neurofibromatosis can be difficult to make in childhood as the disease develops with age. Examination of both parents may make the diagnosis more certain in the child, if one parent is found to be showing signs of the disorder. Sometimes genetic conditions can vary in severity from generation to generation and so the parent may not have realised that they have the

same condition as the child. This will generate an Episode of Care for the parents even though they were not referred. The examination will need to be recorded and the consequences communicated to the patient and their GP. Follow- up will need to be instigated for the child and parent affected.

If the responsibility of care is to the family rather than the individual, as is the case in genetic medicine, the diagnosis of a genetic condition will have consequences to other family members and will generate potential referrals if other family members want to take it further.

> ***Example 3: A patient is diagnosed by a cardiologist with 'the long QT syndrome', which predisposes to sudden death. The first-degree relatives are now all at-risk and need to be seen and tested. A referral is made to the Clinical Genetics Service***

This is usually accomplished through the family. The clinical genetics team will contact the index case and through them offer the family members, identified from the pedigree to be at-risk, information about a genetic referral. Genetic testing for long QT will enable those who are at-risk to be distinguished from those who are not at-risk. One referral can lead to many 'Episodes of Care' for different relatives. Usually relatives are seen in different centres around the country depending on where they live.

2.4.2 Clinical Genetics recommendations for variability in interpretation

There will be differing professional judgements as to what constitutes a separate 'Episode of Care' for another family member and what constitutes normal discussion with relatives of the index case. This could cause differences in costs charged for the service. In normal clinical genetics practice, when a child is referred with developmental delay, it is usual to take a family history and ask about the health of the parents in case it is of relevance to the problem in the child. However, this may be purely related to the health of the child. For example, a child referred with possible Down's syndrome on the post- natal ward will require a history and examination and after the assessment a discussion with the parents specifically related to the health of the child. Talking to the parents of the child about their child does not constitute a separate 'Episode of Care' for the parents.

However, if it transpires during the consultation, that a parent has specific health needs related to the diagnosis in the child, then an 'Episode of Care' for one or both parents may be warranted. For example, in the above example the parents may ask to be seen again to discuss the risks for themselves of having another child with Down's syndrome and prenatal options. Significant time and communication will thus be required for the parent(s) separate to the needs of the child. The Do

Once and Share Clinical Genetics team recommend that an 'Episode of Care' is included for a relative if relevant genetic advice is given. This usually means the consultation with the relative, as well as the index case, warrants separate or additional communication to a health professional concerning that individual.

2.4.3 Family History information and personal pedigree

All health professionals draw pedigrees and use family history when making a diagnosis and assessing risks of disease, but this is of particular relevance to the practice of genetic medicine, as so many of the disorders in question are due to genes that play a significant part in the susceptibility to disease. Therefore, family history is a tool used extensively in genetics.

Family history taking allows the use of family information to aid diagnosis and assess risk for another member of the family. It can be used retrospectively or proactively;

> ***Father died of a heart attack at 35 years. Whilst running for the bus his son, now aged 34 years, suffers from central chest pain. He wants to know what is the likely diagnosis?***
>
> ***Mother has Huntington's disease. Her son wants to know what is his risk of developing the disease?***

In both examples, the use of family information is of relevance to the diagnosis, or potential diagnosis, of relatives. Ideally, there should be a generic tool for all patients looked after in the NHS to allow people to record their own family history information. This is akin to the US Surgeon General's initiative on family history (http://www.hhs.gov/familyhistory/). Individuals can be considered to own this information given that they supply it, and even though this pedigree will contain information about relatives, this is not seen as an ethical dilemma. This point has been extensively debated by the Consent and Consanguinity committee of the Joint Committee of Medical Genetics (Consent and Confidentiality, Royal College and Physicians 2006 unpublished) and the principle established.

It should be noted that some pedigrees are complex, with multiple consanguinity and/or multiple partners and so the IT tool needs to be sophisticated. This report does not include the dataset for the personal pedigree information record, as this requires further consultation.

Within the Do Once and Share Clinical Genetics care pathway, the difference between a personal pedigree based on personal knowledge of the health of relatives and a modified genetics pedigree, which has resulted in accessing health records of relatives to confirm disease status and inform risks, has been identified. Every patient seen in clinical genetics will have a modified family pedigree. It is initially derived from the information known by the patient. Through informed consent, the pedigree is verified where key issues, such as a genetic test result, histology or a death certificate, are required. Therefore, the notes of patient A, who is referred with breast cancer, will have information on her mother and father, her brothers and sisters, and perhaps her second-degree relatives. This information will include not only personal details but details of illnesses, and relevant medical reports.

2.5 Referral triage

At present, there are few guidelines for appropriate referral to clinical genetics services except for some cancer referrals. These need to be developed and will be a key component in identifying touch points between the clinical genetics service and other NHS Care Pathways. Current practice involves scrutiny of referrals before accepting the referral by the service and this is denoted in the pathway as 'Referral Triage'. In addition, clinical genetics services offer specialised clinics. Some of them are in conjunction with other specialists - so called 'joint clinics' and there is sub specialisation within genetics, such that some health professionals have specific expertise within the service. As a result, most referrals are assessed to identify the most appropriate clinic for them to attend, both in terms of expert staff and nearness to the family's home. Clinical genetics is therefore not immediately amenable to the standard Choose and Book arrangements. Work is in progress to establish how to implement this in the service.

2.5.1 Clinical Genetics DOAS recommendations for variation in interpretation

Referral guidelines are required for referral to clinical genetics services.

2.6 Coding of diagnosis in Clinical Genetics

A survey of centres within the clinical genetics community has established that all centres are using different codes to represent different diagnoses and issues under discussion at consultation. This is a major drawback to sharing information across centres and must be overcome. Other countries have a national approach; in Holland all clinical genetics centres accept a national coding system (AJMG ,70;4;444-447, 1998). The Department of Health have recognised this problem in England and are

coordinating work with SNOMED - CT. It must be recognised, that there are more than 4000 single gene disorders and many conditions that have not yet been coded. ICD and OMIM (on line Mendelian Inheritance in Man) codes have not as yet met this challenge or were created for different purposes. It is recognised that a coding system that is based on a hierarchical root and branch structure, such that disorders that are linked are grouped, has advantages but any agreed coding system would have advantages over the present position. The Dutch system recognises the difference between a definite diagnosis and a possible diagnosis and non-single-gene issues such as consanguinity.

The importance of deciding on a coding system is emphasised when one considers the power of electronic records in facilitating the gathering of data recorded in the patient notes, particularly for rare disorders.

2.6.1 Clinical Genetics DOAS recommendations for Clinical Genetics coding

The DOAS Clinical Genetics project suggests that a coding system is decided on for England and all clinical genetics centres agree to use it. It must allow for the thousands of genetic conditions and the issues that do not have a single gene cause. Funding will be required to maintain any coding system, as new diagnoses will continue to be discovered. The code needs to be the same as that used in the laboratory genetics services.

2.7 Minimal clinical data set

The entry by a clinician of a diagnostic code within the electronic notes, in the future, could allow for the collection of a minimal clinical data set for each coded disorder. This would be a powerful way of routinely collecting vital natural history data on rare disorders.

For example, the entry of the code for Marfan syndrome could automatically bring up the management pathway for the condition. This will include an electronic web based form facilitating the collection of a minimal data set, such that over time the natural history of the condition would be documented because data from clinics all over the country would be collated. In Marfan syndrome, aortic dilatation, age and medication might be included, providing invaluable information on the outcome of the disease. The NHS should use its assets and the gathering of information that is currently lost in the paper notes of each genetics centre is one use of pooled electronic records. At the same time information on the condition from the latest reviews and papers could also be available automatically.

2.8 Codes for ethnic groups

In Clinical Genetics, ethnicity is of particular importance, as it can inform which genetic tests to perform. As a result the codes for ethnic groups are more detailed than in other specialties. Hence Genetic codes on ethnic groups differ from other specialities in this respect.

3.0 Ethical issues of the electronic care record for clinical genetics

3.1 Family notes

In genetic medicine, the family history is recorded in 'family notes'**.** Individuals within the family have individual sections but information from one, relevant for others, can be viewed. Paper family notes exist within all Clinical genetics services and are universally adopted. They need to be maintained for effective management of genetic disorders and notes coexist alongside individual hospital records. The ability to recognise within individual care records, that a set of family notes exists involving that patient, should be available. It is currently organised at local Trust level. Furthermore this information will need to be kept indefinitely as it has lifelong relevance for all descendents.

3.1.1 Benefits of family notes

- Efficient and cost effective practice

The reduction of repetition clearly costs less and does not incur the dangers of incorrect transcription. Information gathering takes time which costs money. If this can be done once for all family members at-risk, rather than repeated for each, then this is cost effective

3.1.2 Risks for genetic medicine without family notes

- Risk of incorrect diagnosis

Genetic tests are often very specific and may be unique within a family. At present techniques are such that screening tests for genetic mutations are not always able to identify all mutations present. If an individual has a known particular mutation within a gene, the genetic laboratory testing a relative can test for this mutation alone and not rely on a less accurate screening test of the whole gene. The prior knowledge of the mutation thus prevents the possibility of missing it in a routine screen of the gene. The diagnostic test in the relative is therefore greatly simplified if the knowledge of the test result in the family member is known.

> ***A female patient has severe developmental delay due to a small unbalanced rearrangement of the chromosomes identified using molecular cytogenetic techniques and***

not visible on routine chromosome analysis. The aunt of the patient is pregnant and wants prenatal diagnosis, as she knows that she is at increased risk of having a child with the same developmental problem as her niece.

If the precise genetic findings in the niece are not appreciated, the aunt may well be offered routine amniocentesis to identify a chromosome rearrangement in her unborn child. This test would miss the potential chromosome rearrangement in the family and is not a useful investigation. Sharing of family information in family notes prevents this risk and would mean that the aunt would be offered appropriate testing.

3.1.3 Current ethical dilemmas revolve around the concerns of the individual versus the family.

- The current concept of the patient record is personal to the patient
- In future the IT spine needs to collect minimal national data for simplicity
- Patients need control of and know where notes are being kept on them in the NHS
- Patients have the right 'to know' and 'not to know' their risks

versus

- Genetic information of an individual is of relevance to biologically (and sometimes socially) related individuals
- Genetic medicine works efficiently with the use of family notes rather than individual notes where genetically relevant information is shared within families
- If clinical geneticists and/or all health professionals involved in genetic medicine, have a legitimate relationship with a relative of the index case then collecting information using electronic records should be easier than it currently is

3.1.4 Consent

Consent can be obtained for each episode of access to the notes of relatives. This most closely equates to current practice. Once the information is obtained it remains indefinitely within the family notes unless there are clear instructions to remove it, which is rarely requested. This approach is not ideal or simple. It means that much relevant information is lost and/or is never available as there are a multitude of reasons why contacting a relative and locating the information from notes is not possible. There are also no consistently applied rules of consent. How long and for

whom does the consent exist when it is given? For example, if doctor C gets consent in 1999 can the information still be used in 2003, by doctor C and/or a member of the new genetic team, for the original index case or another relative?

3.1.5 Emergent themes

- Consent does not differ for electronic records compared to paper records
- The ability to search electronic records will be easier and therefore rules of access need to be regulated
- Patients accept the need for family information sharing
- When and how consent is obtained from the relatives to access information and to be included in family notes, remains an issue

3.1.6 clinical genetics DOAS recommendations about shared family information

Clinical genetics health professionals (and/or health professionals having undergone a genetic education program) have a legitimate relationship with relatives of index cases, for the purposes of genetic medicine. This is akin to the special relationship that exists for GU services. Families recognise that information from family members may be of relevance to other family members. Therefore, a geneticist seeing patient 1 has a legitimate relationship with relative B in order to gather appropriate information.

Access to medical records of relatives is possible for clinical genetics health professionals provided relatives have a) agreed in advance or b) been contacted and asked to consent. In the former model, (a), it could be established within the ground rules of the Electronic Care record (ECR) that patients could either opt to allow their health records to be accessed for the purposes of the health of relatives, with the option of protecting certain information from any access (the opt 'out' model), or they could opt to place certain information within 'family sealed envelopes' (the opt 'in' model). In this latter proposal, NHS health professionals will require considerable education to help patients realise what sort of information can be useful. Identifying what information to share can be difficult to define. While it is straightforward to assume that most will realise that a genetic test result is likely to be considered relevant to other family members, seemingly, non-genetic information can also be of relevance. If patient B had a bowel polyp of histology type X and information is available that his brother, too, had a bowel polyp of type X then it is more likely to be genetic as both polyps have the same histology, even though a histology report may not be considered genetic information. The child/niece of these siblings will have a higher risk of

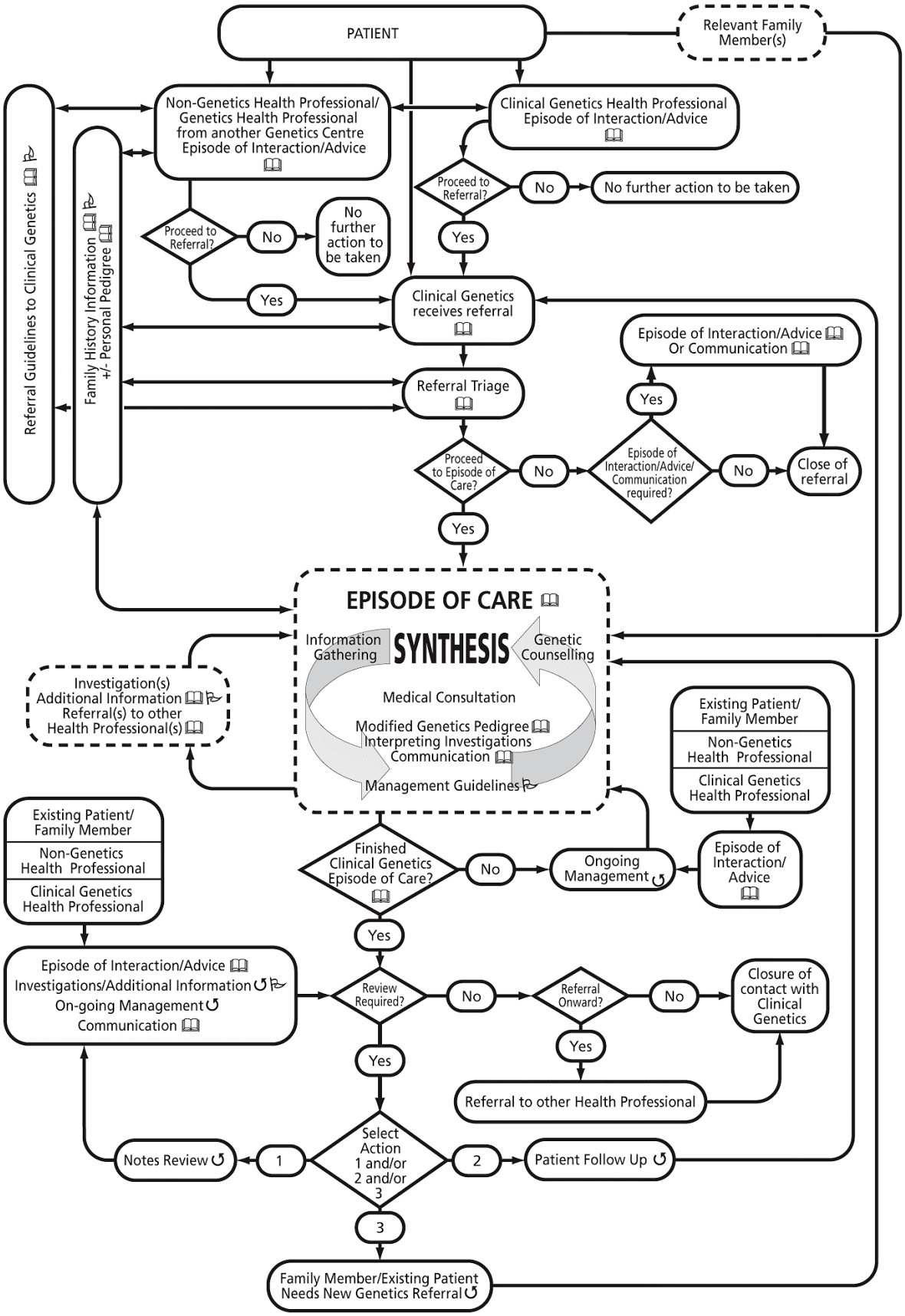
developing polyps and warrant different preventative methods if the polyps are the same histology than if it is shown that they are different. In addition, it needs to be remembered that negative information is also important when calculating risks. For example, knowing that a patient, who had a bowel operation, did not have polyps may also be useful information to at-risk relatives. The 'opt out' model is thus preferred but needs to be openly debated. This could be a topic to discuss with the Human Genetics Commission.

It is suggested that the modified genetics pedigree will sit within the family notes, whereas the personal pedigree may well be relevant to all individual notes. IT help will be required to create electronic family notes for genetics centres. In the interim they will continue to keep paper records to record the details. Family records may well be of relevance to other specialities as subsets of common disorders are identified as being inherited in a Mendelian fashion e.g. for dermatology and ophthalmology.

Include in the spine the family notes number which will denote both the genetic family number and the genetics centre therefore creating the ability to know which genetics centre is keeping the record e.g. SOT123444 (SOT for a Southampton record; 123444 for the genetics family number).

4.0 The Care Pathway for Clinical Genetics

Figure 1: The Care Pathway for Clinical Genetics



5.0 Three phases of the Clinical Genetics Care Pathway

The clinical genetics **care pathway** (Figure 1) follows the patient journey through the Clinical genetics service. It is divided into three distinct phases of clinical activity:

- Pre-referral to referral
- Clinical Genetics Episode of Care
- Review

The care pathway is accompanied by a **dataset.** The dataset consists of dataset items. Each element of the care pathway has a corresponding, numbered dataset item.

5.1 Phase 1 - Pre-referral to referral clinical activity

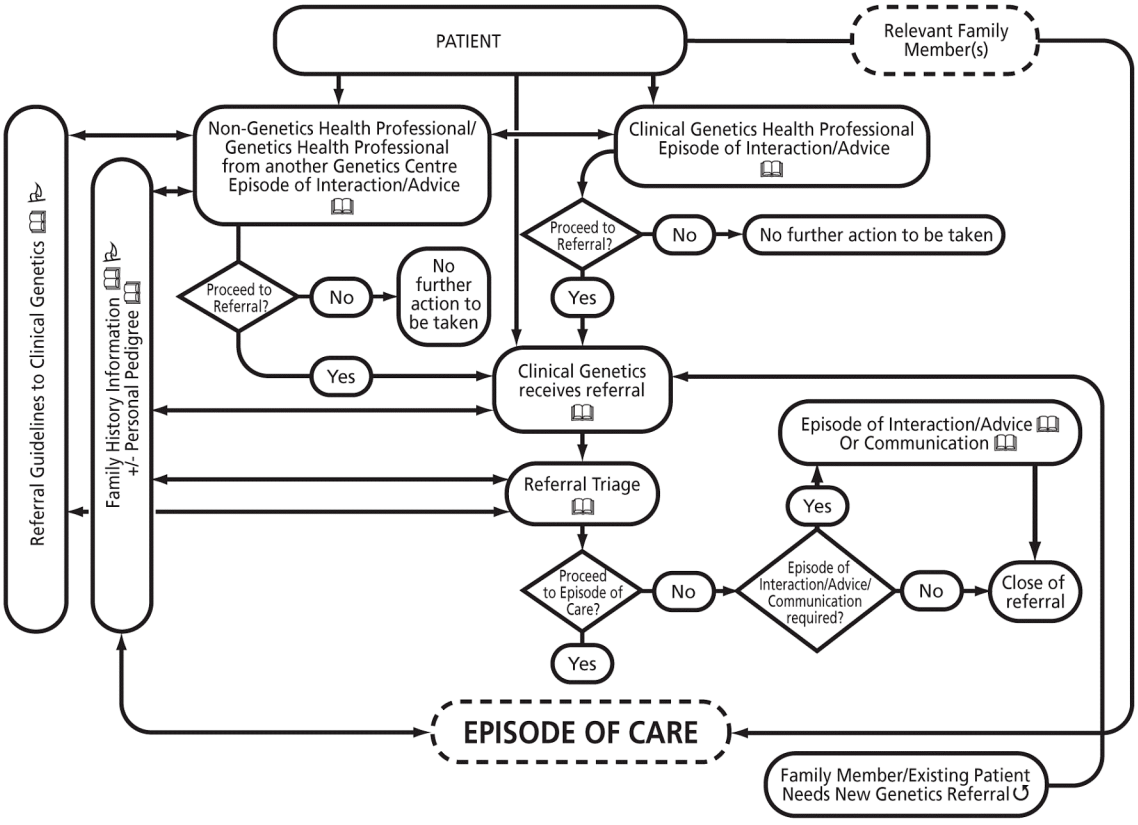


Figure 2: Pre-referral – referral clinical activity

This phase of the pathway identifies clinical activity that occurs before referral for an Episode of Care.

5.1.2 Access to Clinical Genetics Services

The patient is able to access the service by 4 routes:

- Self directed for advice/information
- Self referral
- Via a non-genetics health professional or a genetics health professional from another genetics centre. The health professional may either:
  - Seek advice from clinical genetics or,
  - Refer the patient
- As a family member or an existing patient requiring a new genetics referral

5.1.3 Episode of Interaction or Advice (1.0)

It is usual for Clinical genetics services to receive letters, emails and phone calls from patients and health professionals seeking advice directly about the management of a patient or requesting genetic information before a patient is referred to the service. If the enquiry can be dealt with entirely within this 'two-way episode of communication' this is termed an 'Episode of Interaction/Advice'. The discussion needs to be recorded so that the information discussed can be tracked, should the patient/health professional re-contact the department. There will be occasions when the advice will not be patient related so the dataset includes the ability not to include patient identifiers if not appropriate. This dataset allows Clinical genetics services to record, and therefore capture all the work undertaken before referral. It is also applicable when an episode of interaction or advice occurs at any other point on the pathway. The dataset that accompanies this episode, records key data allowing searching of patient name, dates and issues discussed.

An Episode of Interaction or Advice is defined as:

> ***An occasion when a request for advice is instigated by a patient, a non-genetics health professional or a genetics health professional from another centre. The interaction neither is planned, nor is equivalent to an outpatient appointment.***

Dataset items include:

> ***Advice requested: when (1.1), how (1.2) and by whom (1.3)***
>
> ***Demographics of the patient (1.6)***

Status of the patient (1.6.25). A description of the category of the patient e.g. new patient, not referred, existing patient, family member etc

Code for sequence of Episode of Interaction/Advice (1.7) relating it to the patient

Advice given: by whom (1.8), when (1.9), how (1.10) and time taken (1.11)

Outcome of Episode of Interaction/Advice (1.13). An identifier to determine if advice resulted in referral, follow up appointment, or, a new investigation etc

5.1.4 Family history information (2.0)

It is envisaged that all patients will be requested to forward their family history information, as a pedigree (family tree), to clinical genetics before referral. However, it may also be collected at any point on the care pathway to reflect current practice.

Dataset items include:

Date information requested (2.1) and received (2.2)

At present, there is no consensus on the data items that should be requested within the family pedigree. This is outside the scope of the project.

5.1.5 Clinical Genetics referral (3.0)

A 'new referral' is defined as:

A patient or a family member, who has not previously been referred, or a current patient that requires a new counselling event due to a new issue, or a patient who has not had an outpatient appointment/Significant Contact within the last 12 months (definition taken from GENCAG Quality Markers 2006).

The referral source can be:

- A referral from a non genetics health care professional or geneticist from another genetics centre
- A self referral
- A referral by the Clinical genetics service of a family member identified as a result of an existing family member

Dataset items include:

The date of the decision to refer (3.1)

How the referral was made (3.2) and by whom (3.3)

Priority type (3.6) and referral issue(s)/diagnosis(es) (3.7) as stated on referral

Demographics of patient (3.8 as 1.6)

Date referral received (3.9)

Referred to whom (3.10)

5.1.6 The referral triage (4.0)

All referrals received into Clinical genetics services are triaged by the clinical genetics team and decisions are made regarding the management that should follow.

Dataset items include:

The code of the person who assessed the referral e.g. genetic counsellor, consultant geneticist, specialist registrar (4.1)

Priority type as assessed by clinical genetics (4.3)

Meets referral guidelines (as appropriate) (4.4)

Decision to proceed to Episode of Care (4.5)

Reason to reject referral (as appropriate) (4.5.1)

Communication from Clinical Genetics Department (4.6). (N.B. This dataset is used for all episodes of communication that occur when a referral has been received, during and after the Episode of Care). It includes:

Communication to whom (4.6.1)

How communicated (4.6.2)

Date communication sent (4.6.3)

Reason for delay (if appropriate) (4.6.4)

5.2 Phase 2 – Clinical Genetics Episode of Care (5.0)

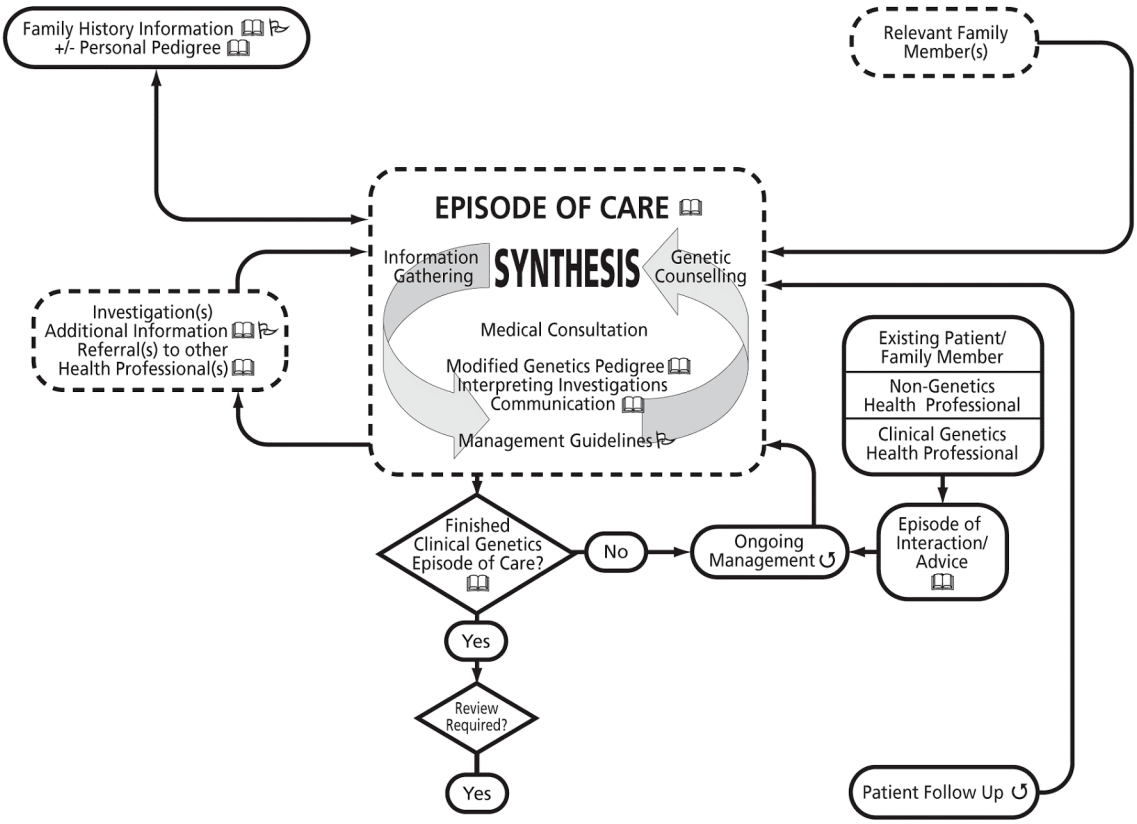


Figure 3: Episode of Care clinical activity

5.2.1 Definition of an Clinical Genetics Episode of Care

A Clinical Genetics Episode of Care is defined as:

> ***An outpatient appointment or several appointments (or the equivalent, termed a planned Significant Contact which takes place in another setting) that is/are required to answer the patient referral reason.***

An outpatient appointment is defined as a planned face-to-face meeting between a patient and genetics health professional in hospital or GP surgery. In clinical practice, it may be replaced by other types of 'Significant Contact' including:

- Face to face contacts at home, on the ward
- Letters, phone contacts, electronic contacts that replace a face-to-face clinic contact

But excluding, for the purposes of this definition:

- Contacts not replacing an outpatient appointment

5.2.2 Clinical components of an Episode of Care

An Episode of Care has three major clinical components and it may require any combination of the following:

- Information gathering
- Genetic counselling
- Medical consultation

The Clinical genetics service may legitimately offer Episodes of Care to relatives not initially referred because of the nature of genetic medicine. Most Episodes of Care result in communication to other health professionals looking after the patient and this can be used to decide if an Episode of Care on a relative is warranted (see the discussion within the introduction section).

Dataset items include:

Waiting times from referral to first Significant Contact (5.1)

Clinical case discussion (5.2)

Outpatient appointment/Significant Contact details (5.3)

Demographics for each patient receiving an 'Episode of Care' (5.4. as 1.6)

Personal pedigree (5.5)

Modified genetics pedigree (5.6). At present, there is no dataset for the items to be collected other than the health professional recording it

Issue(s)/diagnosis(es) under investigation (5.7)

Investigation/additional information type requested (5.8)

Communication following Significant Contact within Episode of Care (5.9)

5.2.3 Finished Clinical Genetics Episode of Care (6.0)

More than one outpatient/Significant Contact with different members of the genetics team may be necessary before the Episode of Care is termed 'Finished' (6.1). The decision that the Episode of Care is 'Finished' is a professional one but is broadly related to answering the referral issue. At the end of an outpatient appointment (Significant Contact) the genetics health professional must ask the question 'is the referral question answered as far as possible? Are there outstanding results/investigations? If complete the Episode of Care is termed finished, if not there is on-going management and the Episode of Care is not closed.

The first and subsequent Episodes of Care are coded such that the number of outpatient appointments (Significant Contacts) within them can be recorded alongside the code for the Episode. For example, the first Significant Contact within the first Episode of Care is coded (1/1), the second Significant Contact within the first Episode of Care is coded (1/2), the second Significant Contact within the second Episode of Care is coded (2/2) etc.

It is recognised that there may be contact with patients over and above an outpatient appointment (or equivalent) that is related to finishing the episode and this is captured in two ways:

- Some contacts will be recorded under the 'Communication' dataset section (4.6), when initiated by the clinical genetics department and involving communication to the patient e.g. by letter
- If a patient/non-genetics health professional instigates a request for further Interaction/Advice, while the Episode of Care is still ongoing, and another outpatient appointment/Significant Contact is not deemed necessary, then it may be judged to be a separate 'Episode of Interaction/Advice' (1.0). Coding will allow this to be included within the Episode of Care and counted in addition to the 'Outpatient appointments/Significant Contacts' using nomenclature that distinguishes an outpatient appointment from an episode of advice (e.g. A1, A2, A3 to indicate the sequence of Episodes of Interaction/Advice and 1,2,3 to indicate outpatient appointments)

5.3 Phase 3 - Review clinical activity

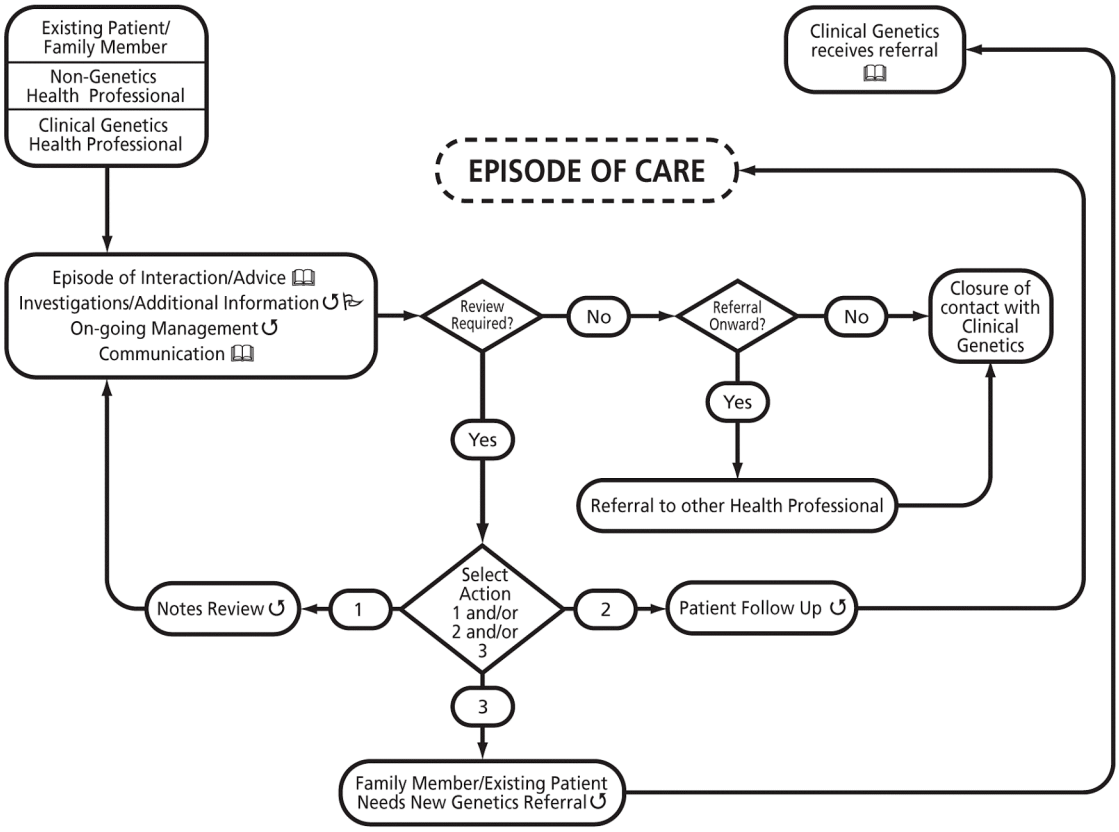


Figure 4: Review clinical activity

Once a decision has been made to finish the Episode of Care, a decision is then required to decide if a review is needed.

There are 4 review options

- No review necessary
- A review of the notes
- Patient follow-up required
- Family member/existing patient needs a new genetics referral

It should be noted that the Episode of Care for one individual may lead to the need for relatives to be referred for clinical genetics input.

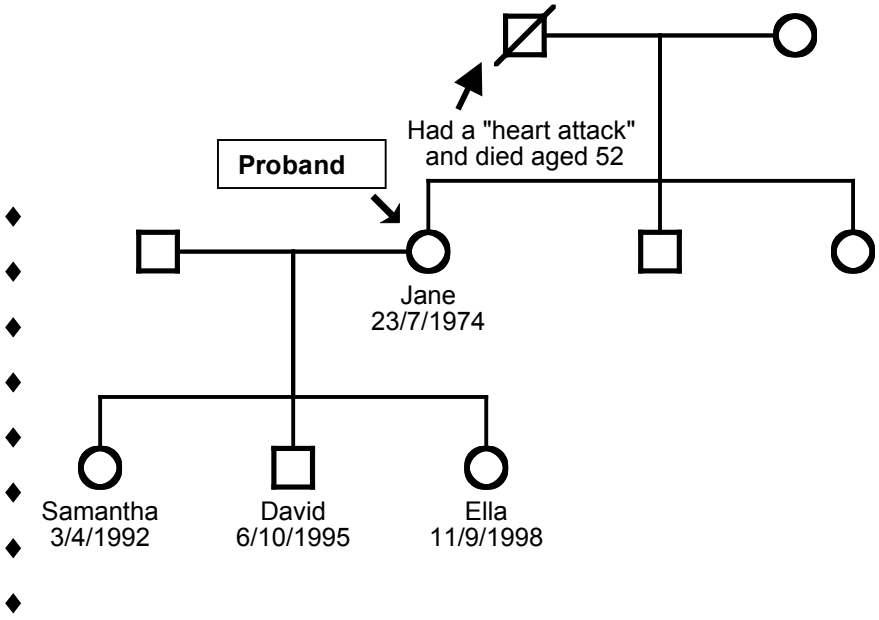
Dataset items include:

Date Episode of Care closed (6.2)

Episode of Care closed by whom (6.3)

Referral onward required (6.8)

5.4 Care Pathway illustrated by case study



Jane, aged 31 years, was admitted to the A&E department with severe chest pain

The diagnosis was a ruptured ascending aortic aneurysm

Following cardiac surgery, she was informed by the cardiac surgeon that the diagnosis was possibly Marfan syndrome

Jane was aware, after reading on the Internet, that there were possible genetic implications for her family

She was concerned for her own health and the risks for her children

She asked her GP for a referral to clinical genetics

Episode of Care 1 & Significant Contact 1

- Jane was seen in clinic by a genetic counsellor
  - Her medical and family history were recorded in the family notes
  - She was referred to an ophthalmologist for a specialist examination to aid diagnosis
  - Request for father's death certificate was sent

Episode of Care 1 & Significant Contact 2

- Jane was seen in clinic by consultant geneticist
  - A medical examination suggested a clinical diagnosis of Marfan syndrome
  - Blood was taken for gene mutation testing
  - Results communicated by letter (***Communication)***
  - Jane referred for lifelong surveillance to cardiologist
  - Father's death certificate confirmed the cause of death as 'ruptured aorta'

This work represented the ***Finished Clinical Genetics Episode of Care***.

- The result highlighted the potential risks to her first degree relatives
- Jane's 3 children, Samantha, David and Ella were offered appointments with the clinical genetics service (***option 3 following review)***
- Jane's brother and sister offered referral to local genetics centre (***communication***)

A new Episode of Care 1 & Significant Contact 1 for each child

- All children were seen in clinic for the first time by genetic counsellor
  - All wanted to be tested and blood testing arranged after appropriate discussion

Episode of Care 1 & Significant Contact 2 for each child

- All children were seen in clinic again by the genetic counsellor to discuss the implications of results
  - Results demonstrated Samantha and David both had inherited the gene mutation in the family for Marfan syndrome
  - Samantha and David referred to a paediatric cardiologist for life long screening
  - Ella discharged from clinical genetics and no review necessary

5.5 Case scenario resumé

Jane's referral resulted in:

- 1 Episode of Care made up of 2 Significant Contacts for Jane
- 1 referral to the ophthalmologist for system examination
- 1 referral to the cardiologist for lifelong surveillance
- 3 referrals to the Service for Jane's children, Samantha, David and Ella
- 1 Episode of Care made up of 2 Significant Contacts for each child
- 2 referrals to paediatric cardiology
- 2 referrals to other Clinical genetics services in the UK
- 1 Episode of Interaction/Advice

6.0. Illustrated Care Pathway: A family history of Marfan syndrome

1. Jane consults with GP because of a possible diagnosis of Marfan syndrome.

2. GP makes referral to Clinical Genetics

3. Referral received and triaged by genetic counsellor and consultant geneticist

Hospital notes confirmed aortic rupture

4. Appointment sent to see genetic counsellor in clinic

5. Appointment with genetic counsellor

Concerns about own health and risks to children
Family history taken

6. Episode of Care not finished

Request for father's death certificate
Jane referred to ophthalmologist
Summary letter sent

7. Episode of Care not finished

Offered second appointment with consultant geneticist for medical consultation
Appointment letter sent

8. Appointment with Consultant Geneticist

Medical examination confirmed signs of Marfan syndrome
Referral onward for medical management with cardiology/joint Marfan clinic
Blood test taken for mutation testing

Episode of Care not finished

9. Episode of Care finished Marfan gene mutation identified
Letter sent confirming result

10. Review required (option 3)

Appointment offered to see children Jane's brother, sister and father's relatives offered Clinical Genetics referral to centres near to home

11. Children seen in clinic by genetic counsellor

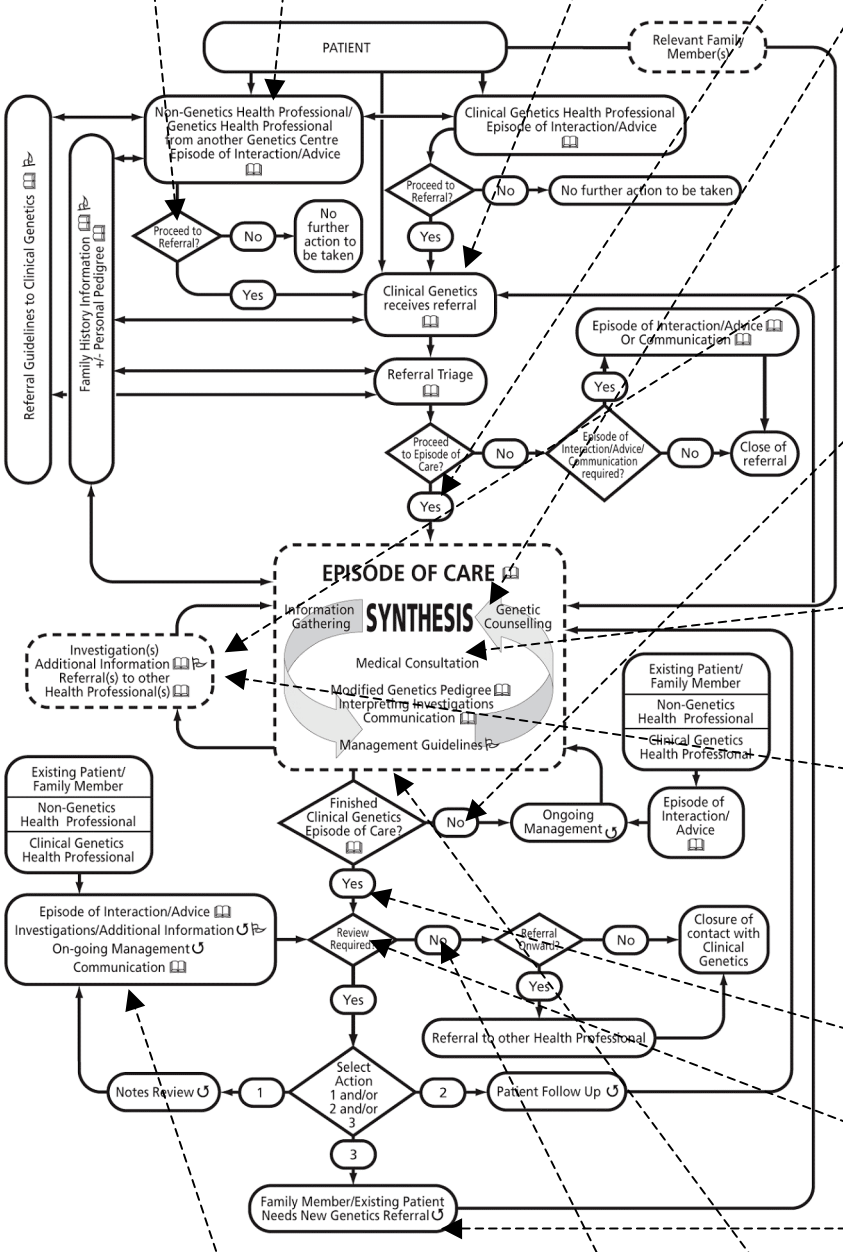
All offered gene testing
Blood taken from all children
Marfan gene mutation identified in eldest daughter and son

11. Review required (no)

2 children referred onward to cardiologist/joint Marfan clinic and ophthalmologist
Ella discharged – no follow-up
Episode(s) of care finished for 3 children
Family asked to re contact Clinical Genetics when children wish to discuss diagnosis

12. Episode of Interaction/Advice

Some time later GP phones to enquire if David can play normal rugby at school.
On call genetic counsellor informs GP that contact sports are not advisable



7.0 Clinical Genetics Dataset

This data set is made up of data items; the description, the reason for collection, the IT codes and classification. Each part of the pathway has an accompanying dataset with a corresponding reference number.

1.0	EPISODE OF INTERACTION/ADVICE WITH CLINICAL GENETICS				
Data ref no.	**Data item**	**Description**	**Purpose**	**Codes and classifications**	**Core data**
1.1	**INTERACTION DATE**	Date Interaction/Advice sought	To establish the date on which the patient/health professional seeks Interaction/Advice	Date format	YES
1.2	**INTERACTION MODE**	Method by which Interaction/Advice sought	To monitor use of resources	Email Post Telephone Fax Face to face Any combination of these e.g. telephone and letter	NO
1.3	**INTERACTION/ ADVICE PRACTITIONER CODE WHO REQUESTED IT**	The code of the person requesting Interaction/Advice e.g. GP Code, Consultant Code, nurse Code	To monitor education needs of community	NHS Data Dictionary Supporting Information, Practitioner Code (8-character code for GPs consultants, nurses. A default code X9999998 can be used where the request is not from an NHS practitioner consultant or GP e.g. enquiry from the public. Not known Not applicable	YES
1.4	**INTERACTION/ ADVICE ORGANISATION CODE OF REQUESTING PRACTITIONER**	Code of the organisation to which the requester belongs	Funding reasons	NHS Data dictionary PCT code Ability to have not applicable	YES
1.5	**INTERACTION/ ADVICE PRACTITIONER SPECIALITY WHO REQUESTED IT**	The specialty from where the practitioner originates	To monitor patterns and influence education.	NHS Data Dictionary e.g. obstetrics, paediatrics To include patient/not applicable	NO

1.6	DEMOGRAPHICS OF THE PERSON (USE FOR EPISODE OF INTERACTION/ADVICE, REFERRAL OR EPISODE OF

	CARE AS REQUIRED)				
1.6.2	**PERSON FAMILY IDENTIFIER (Genetics file number)**	The clinical genetics family number to which the patient is part of	To link family members to same family	Genetics 12 character number starting with Genetics centre code and then genetics family code Needs code here Including the ability for not applicable	YES
1.6.3	**PERSON FAMILY NAME**	The patient's family name	Required until all have own unique NHS Number or where NHS Number is not available. Required to be used to link other family members previously seen with referred patient. For retrieval of patient details	NHS Date Dictionary	YES
1.6.4	**PERSON PREVIOUS FAMILY NAME**	The patient's previous family name	Required to be used to link other family members previously seen with referred patient. For retrieval of patient details	NHS Data dictionary Including the ability for not applicable	YES
1.6.5	**PERSON FAMILY NAME AT BIRTH**	The patient's family name at birth. Equivalent to maiden name where applicable	Required to be used to link other family members previously seen with referred patient. For retrieval of patient details	NHS Data Dictionary Including the ability for not applicable	YES
1.6.6	**PERSON FORENAME OR PERSONAL NAME**	The patient's forenames	Required to be used to link other family members previously seen with referred patient. For retrieval of patient details	NHS Data Dictionary	YES

1.6.7	**PERSON PREVIOUS FORENAME OR PERSONAL NAME**	The patient's previous forenames	Required until all have own unique NHS Number or where NHS Number is not available. Required to be used to link other family members previously seen with referred patient. For retrieval of patient details	NHS Data Dictionary	YES
1.6 8	**PERSON USUAL ADDRESS**	Patient's usual address	The address is used to enable analysis by locality. To enable analysis by locality	NHS Data Dictionary	YES
1.6.9	**PERSON POSTCODE OF USUAL ADDRESS**	Patient's usual Postcode	To derive and/or verify Postcode To enable analysis by locality	NHS Data Dictionary	YES
1.6.10	**PERSON PREVIOUS ADDRESSES**	Patient's previous addresses where patient lived	The address is used to enable analysis by locality. To enable analysis by locality	NHS Data Dictionary	YES
1.6.11	**PERSON POSTCODE OF PREVIOUS ADDRESSES**	Patient's previous Postcode	To derive and/or verify Postcode To enable analysis by locality	NHS Data Dictionary	YES
1.6.12	**PERSON CORRESPONDENCE ADDRESS**	Patient's preferred correspondence address	The patient's preferred correspondence address for use for communication	NHS Data Dictionary	YES
1.6.13	**PERSON PREFERRED COMMUNICATION CONTACT METHOD**	A choice of letter, telephone, email	To take into account the patient's wishes as to how they would prefer to be contacted through the process. It could be one mode or many	NHS Data dictionary	YES
1.6.14	**PERSON TELEPHONE (S)**	Patient's phone numbers indicating home/work/mobile	To contact patient if necessary	Number code NHS Data Dictionary	YES
1.6.15	**PERSON EMAIL ADDRESS**	Patient's email address	To contact patient if necessary	NHS Data Dictionary	YES
1.6.16	**PERSON BIRTH DATE**	The patient's date of birth	To enable analysis by age	NHS Data Dictionary	YES
1.6.17	**INTERPRETER REQUIRED INDICATOR**	Identifies patient's need to be accompanied by an interpreter	To enable prior or future arrangement of interpretation services if patient does not converse well or understand English sufficiently to benefit from the Interaction/Advice/ Episode of Care	Yes No	YES
1.6.18	**LANGUAGE**	The patient's language	First language in which patient prefers to conduct the consultation	NHS Data dictionary Language	YES

1.6.19	**PERSON SPECIAL NEEDS (E.G. BLIND, HEARING LOSS, PHYSICAL DISABILITY/ SIGN LANGUAGE)**	The patient's special needs requirement	Identifies any special needs of which clinical genetics need to take account when arranging appointments and facilities	NHS Data dictionary List needed FREE TEXT	YES
1.6.20	**ETHNIC BACKGROUND**	The ethnic background of the patient, as specified by the patient	To enable analysis by ethnicity	**White** British A Irish B Any other white background C **Mixed** White and Black Caribbean D White and Black African E White and Asian F Any other mixed background G **Asian or Asian British** Indian H Pakistani J Bangladeshi K Any other Asian background L **Black or Black British** Caribbean M African N Any other black background P **Other Ethnic Groups** Chinese R Any other ethnic group S **Not stated** Not stated Z FREE TEXT	YES
1.6.21	**ADDITIONAL RELEVANT ANCESTRY**	Record other relevant ancestry e.g. Ashkenazi Jewish	For clinical information to aid genetic diagnosis	Detailed ethnic category (ONS 2001) www.gig.org.uk/docs/workshop_ehinicity_clinical_genetics.pdf	YES
1.6.22	**PERSON CODE OF REGISTERED GP**	The GP with whom the patient is registered	To enable analysis by GP Code	NHS Data Dictionary Supporting Information, Practitioner Code Including none and not applicable	YES
1.6.23	**PERSON CODE OF GP PRACTICE WHERE REGISTERED**	Practice with whom the patient is registered	To enable analysis by GP Practice Code	NHS Data Dictionary Supporting Information, Practice (6 character code) Including not applicable	YES

1.6.24	ORGANISATION CODE OF RESPONSIBLE PCT/OTHER INSTITUTION	PCT or other institution corresponding to the GP Practice Code	To enable analysis by GP PCT Code or other	NHS Data Dictionary Supporting Information, 5 character code starting with '4', allocated by PPA). Note: this can be derived from GP Practice Code.	YES
1.6.25	PERSON STATUS	A description of the category of the patient e.g. new patient, not referred, existing patient etc N.B. Not all patients who are seen/discussed have been referred to clinical genetics. Some are relevant family members who have accompanied index case but have received genetic information/genetic consultation	To analyze patterns of workload. By distinguishing between a referred person and a non-referred person and between first and subsequent Episodes of care the waiting times can be established for individual appointments	01 –New patient, Interaction/Advice only 02 – Existing patient, Interaction/Advice only 03 - Family member of existing family, not personally referred, Interaction/Advice only 04 - Referred index patient, first Episode of Care 05 - Existing patient – subsequent Episodes of Care 06 - Family member but not personally referred – first Episode of Care 07 – Family member of existing family not personally referred– subsequent Episodes of Care 09 other FREE TEXT 010- not applicable N.B. this code needs to link in with the person in question **Needs a code**	YES
1.7	**INTERACTION/ ADVICE CODING EPISODES RELATING TO PATIENT**	To code sequence of Episode of Interaction/Advice with genetics department relating to patient	To identify each Episode of Interaction/Advice and use coding to bind together. Allows ability to separate first Episode of Interaction/Advice from following Episodes of Interaction/Advice	A1/NHS number-First Episode of Interaction/Advice A2/NHS number-Second Episode of Interaction/Advice N.B. The code needs to be able to link the Episode of Interaction/Advice with the patient demographics, and if necessary with the Episode of Care e.g. 1/A1/NHS number	NO

1.8	**INTERATION/ ADVICE CODING PRACTITIONER WHO DEALT WITH INTERACTION/ ADVICE GIVING**	The person who dealt with patient Interaction/Advice	To monitor the skill mix of the clinical genetics team who dealt with the Interaction/Advice requests. For workload and case mix analysis	NHS Data Dictionary Supporting Information, Practitioner Code Assuming designation e.g. nurse, registrar, consultant will be denoted in the code	YES
1.9	**INTERACTION/ ADVICE DATE**	Date Interaction/Advice given	To establish the date clinical genetics service Interaction/Advice sought	Date format	YES
1.10	**INTERACTION/ ADVICE MODE**	Method by which Interaction/Advice given	To monitor use of resources	Email Post Telephone Fax Face to face And any combination of the following	NO
1.11	**INTERACTION/ ADVICE DURATON HOURS (Length Of Time Taken For Interaction/Advice Giving)**	Record time taken for Interaction/Advice session	To monitor use of resources	Duration hours	YES
1.12	**INTERACTION/ ADVICE ISSUE (S) UNDER DISCUSSION**	Record issue discussed e.g. risk of Marfan syndrome or maternal age	To monitor education needs of community/and case mix of clinical genetics departments	Issues codes Problem based codes FREE TEXT	NO
1.13	**INTERACTION/ ADVICE INTENDED OUTCOME**	Specify whether advice resulted in referral/follow up appointment/new investigations	To gauge work load	01 – Referral to department 02 - Follow-up to department 03 – Modification of genetics pedigree 04 – Investigations/additional information 05 – Part of an ongoing Episode of Care Any combinations of the above Other 00 - No further action	NO
2.0	**FAMILY HISTORY INFORMATION**				
Data ref no.	**Data item**	**Description**	**Purpose**	**Codes and classifications**	**Core data**
2.1	**DATE FAMILY HISTORY INFORMATION REQUEST SENT**	The date the information request was sent out	To establish how long it takes to get back the family history (FH) information	Date format Not sent Not applicable Patient initiated	NO

2.2	**DATE RETURNED TO CLINICAL GENETICS DEPARTMENT**	The date the family history information was returned to the department	To establish the date the FH information was returned to the department To establish if FH information was sent with referral letter To monitor waiting times	Date format Not applicable	NO
3.0	**CLINICAL GENETICS REFERRAL**				
3.1	**DATE OF DECISION TO REFER TO CLINICAL GENETICS**	The date on which the referral was made: Date on the letter/fax/ e-mail from referring GP or other hospital department Date of the telephone call from referring GP or other hospital department	To establish the date on which the referring clinician first initiates referral to clinical genetics To identify length of time in the handling of referrals	Date format	YES
3.2	**MODE OF REFERRAL**	The mode of the referral	To identify how referrals are made to clinical genetics	NHS Data Dictionary Supporting Information 01 – letter 02 – email 03 – telephone 04- fax	NO
3.3	**REFERRER CODE**	The code of the person referring the patient e.g. GP Code, Consultant Code, Nurse Code	To identify patterns of referral	NHS Data Dictionary Supporting Information, Practitioner Code (8-character code GPs, 8-character code for consultants, code for nurses. A default code X9999998 can be used where the referral is not from a consultant or GP. N.B. Need a code for self-referral	YES
3.4	**RERRERAL SPECIALITY**	The specialty from where the referral originates	To monitor referral patterns and influence education	NHS Data Dictionary E.g. obstetrics, paediatics Including not applicable	YES
3.5	**REFERRING ORGANISATION CODE**	The organisation at which the person referring the patient is based e.g. the hospital or site's organisation code, the GP's Practice Code	To identify patterns of referral. May be related to funding	NHS Data Dictionary Supporting Information, NHS Trust Site (5 character code) or GP Practice (6 character code)	YES
3.6	**PRIORITY TYPE AS DETERMINED BY REFERRER**	To identify if referrer has prioritised the referral	For analysis of waiting times and comparison between priority determined by assessor	01 -Urgent referral 02 - Soon referral 03 - Routine referral	YES

3.7	**REFERRRAL ISSUE (S)/ DIAGNOSIS (S) CODES**	The main issues/diagnoses for which the patient was referred	To establish the numbers of various issues referred	Issues/diagnoses at referral N.B may be more than one FREE TEXT Need coding	NO
3.8	**DEMOGRAPHICS OF PATIENT REFERRED (AS PER SECTION 1.6)**				
3.9	**REFERRAL REQUEST RECEIVED DATE**	The date that the referral request is received by the clinical genetics department: Date when letter/fax/electronic form is received Date of verbal request	To establish the start date for the referral. To identify length or delay in handling referrals	Date format	YES
3.10	**PRACTITIONER CODE REFERRED TO**	The person to whom the referral is made	To monitor the proportion of referrals made to members of the clinical genetics team	NHS Data Dictionary Practitioner Code	YES
4.0	**CLINICAL GENETICS REFERRAL TRIAGE**				
4.1	**PRACTITIONER CODE ASSESSED BY**	The person who assessed the referral	To monitor the proportion of the assessments made by different members of the multidisciplinary team	NHS Data Dictionary Supporting Information, Practitioner Code 01 Doctor 02 Genetic counsellor 03 Junior doctor 04 Trainee genetic counsellor 05 Administrator Any combination of the above	YES
4.2	**ORGANISATION CODE (CODE OF PROVIDER) ASSESSING REFERRAL**	The organization code of the department providing the referral assessment	To enable analysis by Provider Code	NHS Data Dictionary Supporting Information, Administrative Codes, NHS Trust	YES
4.3	**PRIORITY TYPE AS DETERMINED BY ASSESSOR**	To identify priority of referral	For analysis of waiting times and comparison between priority determined by referrer (see 4.5)	01 -Urgent referral 02 - Soon referral 03 - Routine referral	YES
4.4	**MEETS REFERRAL GUIDELINES FOR AT LEAST ONE ISSUE**	Referral meets referral guidelines	To determine if referral meets referral guidelines	01 –Yes 02 – No Not appropriate	NO

4.5	**PROCEED TO EPISODE OF CARE**	Assessor accepts or rejects referral. (This may be independent of whether or not the referral meets referral guidelines)	To record the number of all referrals accepted and rejected	01 – Yes 02 - No	YES
4.5.1	**REJECT REASON**	Record reasons why referral has been rejected	To determine how referrals are inappropriate	FREE TEXT Not applicable	YES
4.6	**COMMUNICATION FROM THE GENETICS DEPARTMENT (Repeat for each communication episode, before, during and after an Episode of care)**				
4.6.1	**COMMUNICATION TO WHOM**	Record who received communication from the genetics department	For audit and standards	01 – Patient 02 – Family members 03 - GP 04 – Referrer Any combination of the above NHS Data Dictionary	YES
4.6.2	**COMMUNICATION MODE**	The mode of communication	For analysis of the mode of the communication	NHS Data Dictionary Supporting Information 01-letter 02- email 03 -telephone 04- fax Plus ability for multiple combinations of the above	NO
4.6.3	**DATE COMMUNICATION SENT**	The date of the communication	For analysis of the wait to contact	Date format	YES
4.6.4	**DELAY REASON TO COMMUNICATION**	This is a coded data item that can be used to record why there is a wait longer than target times	For analysis of waiting times	03 - Administrative delay (e.g. lost referral, inappropriate to contact patient etc) 99 -Other Not applicable	YES
4.6.5	**DELAY REASON COMMENT**	This is a free text item that must be completed to inform the return on reasons why the existing standard was breached (after any adjustments have been made	To record the actual reason why communication not sent within target.	Free text	NO

5.0	**FINISHED CLINICAL GENETICS EPISODE OF CARE (FCGE)**				
5.1	**WAITING TIMES FROM REFERRAL TO FIRST SIGNIFICANT CONACT WITH GENETICS DEPARTMENT WITHIN A CLINICAL GENETICS EPISODE OF CARE: N.B. ONLY RELEVANT FOR REFERRED INDIVIDUALS (Time in days from receipt of referral until date of Significant Contact within first Episode of Care**				
5.1.1	**DURATION DAYS**	Referral date to first recorded Significant Contact with the genetics department within a clinical genetics Episode of Care	For waiting times	For a referred individual only (Date of referral) – (Date of first Significant Contact within first Episode of Care)	YES
5.1.2	**WAITING TIME ADJUSTMENT**	Record here the number of days that should be removed from the recorded waiting time between referral receipt and date of first Significant Contact within the FCGE. Adjustments are allowed for: A patient who cancels an appointment- clock restarts from the date of the cancelled appointment. A patient Does Not Attend an appointment – clock stops When a patient refuses an appointment within 13 week wait – clock restarts from when patient receives appointment	For analysis of waiting times	Numeric	YES
5.1.3	**DELAY REASON REFERRAL TO FIRST SIGNIFICANT CONTACT WITHIN THE FCGE**	This is a coded data item that can be used to record why patients wait longer than target times	For analysis of waiting times	03 - Administrative delay (e.g. failed to be rebook after DNA, lost referral, etc) Patient decision 99 -Other 88 not applicable	YES

5.1.4	**DELAY REASON COMMENT**	This is a free text item that must be completed to inform the return on reasons why the existing standard was breached (after any adjustments have been made)	To record the actual reason why a patient was not seen within the target	FREE TEXT	NO
5.2	**CLINICAL GENETICS CASE DISCUSSION (Repeat for each case discussion)**				
5.2.1	**DATE OF CLINICAL GENETICS CASE DISCUSSION**	The date patient reviewed	To record the date of the review	Date format Not appropriate	NO
5.2.2	**TYPE OF DISCUSSION**	A description to identify whether the discussion was within the department of at an external meeting/ review	As an audit trail of steps leading to clinical decisions	01 – internal (clinical genetics) 02 – external 03 – internal MDT meeting Any combination of the above None Not appropriate	NO
5.3	**OUTPATIENT APPOINTMENT/PLANNED SIGNIFICANT CONTACT WITHIN AN EPISODE OF CARE**				
5.3.1	**CODING EPISODES OF CARE AND THE SIGNIFICANT CONTACTS WITHIN THEM**	To code sequence of Significant Contacts within the Episodes of Care with genetics department (outpatients equivalent) making up a Finished clinical genetics Episode of Care and subsequent follow up Episodes of Care	To identify each Significant Contact within Episode of Contact (equivalent to an outpatients) and use coding to bind together components of the Finished Clinical Genetics Episode of Care. Allows ability to separate first Episode of Care from following Episodes of Care if that is necessary for funding and waiting time details	1/1- First Significant Contact within first Episode of Care 2/1 - Second Significant Contact within first Episode of Care 3/2- Third Significant Contact within second Episode of Care	YES
5.3.2	**DATE OF SIGNIFICANT CONTACT (WITHIN EPISODE OF CARE)**	The date of the significant contact within the Episode of Care	For analysis of the wait from referral to episode of care	Date format	YES
5.3.3	**PLANNED START TIME OF THE SIGNIFICANT CONTACT (WITHIN EPISODE OF CARE)**	The time the Significant Contact within Episode of Care was planned to start	For analysis of the length of time spent during the Significant Contact within the Episode of Care	Time format	YES

5.3.4	**ACTUAL START TIME OF SIGNIFICANT CONTACT (WITHIN EPISODE OF CARE)**	The actual time the Significant Contact within an Episode of Care started	For analysis of any delay in waiting for the start of the Significant Contact within an Episode of Care against scheduled time of start	Time format including ability to include "did not show"	YES
5.3.5	**END TIME OF SIGNIFICANT CONTACT (WITHIN EPISODE OF CARE)**	The time the Significant Contact within an Episode of Care was complete	For analysis of the length of time spent during a Significant Contact within an Episode of Care	Time format Not applicable	YES
5.3.6	**SIGNIFICANT CONTACT LOCATION (WITHIN EPISODE OF CARE)**	The location where Episode of Care took place e.g. The code for a clinic, code for a home visit, code for a telephone in the department	To identify where care episodes take place. N.B. The code may denote a provider that provides the location of the Episode of Care that differs from the actual provider of the service. Many genetics centres have a hub and spoke model of delivery of care	Clinic code including Multi Specialty clinics Home visit Department Other (Need a list)	YES
5.3.7	**ORGANISATION CODE PROVIDING THE CARE (CODE OF PROVIDER)**	The organisation code of the department providing the care to the patient. N.B. This is not necessarily the same as the code for the clinic location	To enable analysis by Provider Code	NHS Data Dictionary Supporting Information, Administrative Codes, NHS Trust	YES
5.3.8	**MODE OF SIGNIFICANT CONTACT (WITHIN EPISODE OF CARE)**	Mode of Significant Contact within Episode of Care /consultation	To record how the Episode of Care was conducted for work load analysis	NHS Data Dictionary Supporting Information Face to face Telephone Letter Other	YES
5.3.9	**PRACTITIONER CODE RESPONSIBLE FOR SIGNIFICANT CONTACT (WITHIN EPISODE OF CARE)**	The person who has responsibility for the overall care of the patient within the Episode of Care. This may be different from the practitioner who dealt with the Significant Contact	To monitor the proportion of referrals individual members of the clinical genetics team are responsible for	NHS Data Dictionary Supporting Information, Practitioner Code	YES
5.3.10	**PRACTITIONER CODE (S) WHO DEALT WITH SIGNIFICANT CONTACT (WITHIN EPISODE OF**	The person(s) who dealt with patient during Significant Contact within Episode of Care	To monitor the proportion of referrals that individual members of the clinical genetics team deal with within an Episode of Care	NHS Data Dictionary Supporting Information, Practitioner Code	YES

	CARE) **(Repeat for each practitioner who dealt with Episode of Care)**				

5.4	**DEMOGRAPHICS FOR EACH PATIENT RECEIVING EPISODE OF CARE (AS PER SECTION 1.6)**				
5.5	**PERSONAL PEDIGREE**				
5.5.1	**NHS NUMBER AND PERSONAL PEDIGREE VERSION NUMBER CODE**	The NHS number and version number code of the pedigree	To track the version number of each pedigree adaptation	Number format e.g. (Gxxxxxx/1,2,3)	NO
5.5.2	**SOURCE OF FAMILY INFORMATION**	The source of the family information	To identify who gives the family information to draw the pedigree	NHS number/ Name	NO
5.6	**MODIFIED GENETICS (MG) PEDIGREE (Complete for each version of the pedigree)**				
5.6.1	**NHS NUMBER/FAMILY IDENTIFIER AND MG PEDIGREE VERSION NUMBER CODE**	The genetic family number and version number code of the modified pedigree	To track the patients within the pedigree/genetic family and version number of each pedigree adaptation. For maximal search facility need to be able to search on surname, NHS number and family number	Number format e.g. (NHS number/Gxxxxx /1,2,3)	YES
5.6.2	**PRACTITIONER CODE OF THE PERSON WHO MODIFIED THE PEDIGREE**	The person who modified/created the pedigree	To determine the practitioner who records the pedigree information	NHS Data Dictionary Supporting Information, Practitioner Code	YES
5.7	**ISSUE/DIAGNOSIS (S) UNDER INVESTIGATION (Complete for each issue)**				
5.7.1	**ISSUE/ DIAGNOSIS (S)**	The issue/diagnosis code(s) under investigation	To identify disease specific care pathways To compare, retrieve patients, audit and research	Issue/diagnosis code(s) **(Need a list)**	YES
5.7.2	**DISEASE SPECIFIC RECORDED OBSERVATIONS**	The recorded observations for each diagnosis	To allow clinical data to be systematically collected	NHS Data Dictionary Supporting Information	YES
5.7.3	**COMPONENT OF THE**	Identify what aspect of patient care was included in the Significant Contact	For audit and case mix	01 - Information gathering 02 - Clinical	NO

	SIGNIFICANT CONTACT WITHIN EPISODE OF CARE			examination 03 - Formulating differential diagnosis 04 - Genetic information giving 05 - Assessment of risk 06 - Clinical management 07 - Co-coordinating surveillance 08 - Instigating investigation 09 – Interpreting investigations 10 - Additional psychosocial support 11- Research Any combination 12 – Other FREE TEXT	
5.8	**INVESTIGATION/ ADDITIONAL INFORMATION TYPE REQUESTED (Repeat for each investigation/additional information request)**				
5.8.1	**INVESTIGATION/ ADDITIONAL INFORMATION TYPE**	Chromosomes, mutation analysis, Other pathology investigation, Radiology, clinical photography, referral to other specialty for an opinion. Death certificates, medical records, additional family information, radiology, pathology results, a referral onwards for a clinical opinion is included here if the findings will influence management and are important in order to Finish the Episode of Care. This is not for referral onwards to another clinician following FCGE.	To record the type of investigation/additional information type requested	NHS Data Dictionary Supporting Information 01 – chromosomes 01 – sub-microscopic aberrations 02 – Molecular analysis – gene symbol/OMIM number/gene name 03 – other 04 – radiology 05 - referral to other specialty for opinion where result is awaited before close of Episode of Care and code to de-note which specialty FREE TEXT required. (needs a list)	YES
5.8.2	**CONSENT OBTAINED FOR INVESTIGATION/ ADDITIONAL INFORMATION REQUEST**	Consent to investigation/information discussed	To ensure that consent is obtained to carry out investigation	01 – yes 02 – no Not applicable	YES
5.8.3	**DATE INVESTIGATI**	Date that investigation/information was requested	To track the reporting of investigations/ information gathering requested	Date format	YES

	ON /INFORMATIO N REQUESTED				

5.8.4	**DATE INVESTIGATION /INFORMATION REQUESTED**	Date that investigation/information was requested	To track the reporting of investigations/ information gathering requested	Date format	YES
5.8.5	**PRACTITIONER CODE AUTHORISING THE INVESTIGATION/ ADDITIONAL INFORMATION GATHERING**	Personal code of the practitioner who authorised the investigation/additional information	To determine the person who authorised the investigation/additional information gathering	NHS Data Dictionary Supporting Information, Practitioner Code	YES
5.8.6	**ORGANISATION CODE WHERE THE REQUEST FOR INVESTIGATION/ ADDITIONAL INFORMATION IS SENT**	The code of the organisation where the investigation/request for information is sent. This is usually the place where the test is performed. It is the code of the place that is responsible for giving the result/sending information	Required as different activities for a patient may be carried out at different service provider sites. To track and monitor	NHS Data Dictionary Supporting Information, NHS Trust Site (5 character code) or GP Practice (6 character code)	YES
5.8.7	**DATE INVESTIGATION RESULT/ ADDITIONAL INFORMATION IS EXPECTED**	This is the approximate date when the investigation is likely to be available	Tool to track the investigation result	Date format	NO
5.8.8	**DATE INVESTIGATION RESULT/ ADDITIONAL INFORMATION IS RECEIVED**	Date that investigation result received	To track the reporting of investigations requested	Date format	YES
5.8.9	**DELAY REASON TO INVESTIGATION RESULT/ ADDITIONAL INFORMATION RECEIPT**	This is a coded data item that can be used to record why there is a wait longer than target times	For analysis of waiting times for investigations/information requests	03 - Admin delay (e.g. lost test) Laboratory error etc... 99 - Other Not applicable	NO
5.8.10	**DELAY REASON TO INVESTIGATION RESULT/ ADDITIONAL INFORMATION RECEIPT COMMENT**	This is a free text item that must be completed to inform the return on reasons why the existing standard was breached (after any adjustments have been made	To record the actual reason why communication not sent within target	FREE TEXT	NO
5. 9	**COMMUNICATION FOLLOWING SIGNIFICANT CONTACT WITHIN EPISODE OF**				

	CARE (AS PER SECTION 4.6)				
6.0	**FINISHED CLINICAL GENETICS EPISODE OF CARE (MAY CONTAIN SEVERAL SIGNIFICANT CONTACTS)**				
6.1	**FCGE CODE**	Code to link FCGE with Significant Contacts within it	To capture components of the FCGE	Code e.g. F1/1,2,3 Finished first Episode of Care with 3 Significant Contacts	YES

6.2	**DATE EPISODE OF CARE CLOSED**	Date Episode of Care closed	To identify the date Episode of Care closed	Date format	YES
6.3	**PRACTITIONER CODE RESPONSIBLE FOR DECISION FOR FCGE**	The person who has responsibility for closure of the Episode of Care	For case mix information and to identify responsibility	NHS Data Dictionary Supporting Information, Practitioner Code	YES
6.4	**DURATION DAYS TO COMPLETION OF FCGE**	Referral date to Finished clinical genetics Episode of Care (FCGE)	For waiting times	For a referred individual only (Date of referral) – (Date of FCGE)	NO
6.5	**DELAY REASON REFERRAL TO FCGE**	This is a coded data item that can be used to record why patients wait longer than target times	For analysis of waiting times	03 - Administrative delay (e.g. failed to be rebook after DNA, lost referral, etc) Patient decision 99 -Other 88 not applicable	NO
6.6	**DELAY REASON REFERRAL TO FCGE COMMENT**	This is a free text item that must be completed to inform the return on reasons why the existing standard was breached (after any adjustments have been made)	To record the actual reason why a patient was not seen within the target.	FREE TEXT	NO
6.7	**DELAY REASON REFERRAL TO FCGE COMMENT**	This is a free text item that must be completed to inform the return on reasons why the existing standard was breached (after any adjustments have been made)	To record the actual reason why a patient was not seen within the target.	FREE TEXT	NO
6.8	**REFERRAL ONWARD TO OTHER SPECIALITY**	Refer onward to other specialty after close of care episode and when referral onward does not influence outcome for current care episode	This is to record whether clinical genetics refers onward	NHS Data Dictionary Supporting Information 00 - no	YES
6.9	**ISSUES/DIAGNOSIS AT FINISH OF CLINICAL GENETICS EPISODE**				
6.9.1	**CERTAINTY OF DIAGNOSIS IF RELEVANT**	To record how certain is the diagnosis (if applicable)	For information	01 – yes go to 6.7.2 01 – no Not applicable	NO
6.9.2	**BASIS OF THE CERTAINTY OF THE DIAGNOSIS**	This field records how diagnosis was made Choose field most responsible for certainty of diagnosis (if applicable)	To establish how the diagnosis was made for comparison.	01 – clinical 01 family history 03 - chromosomes 04 – molecular analysis 05 Molecular cytogenetics 06 – radiology 07 – pathology 08 diagnosis as referred, 09 not an issue for this care episode FREE TEXT	NO

				Any combination of the above .Not applicable	
7.0	**EPISODE OF INTERACTION/ADVICE (AS PER SECTION 1.0)**				
8.0	**COMMUNICATION (AS PER SECTION 4.6)**				