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University Healthcare
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Subject: Guidance for referring to the Royal Devon Clinical Genetics service

Dear colleagues,

In October 2024, responsibility for the delivery of Clinical Genetics services for Somerset transferred to the Peninsula Clinical Genetics at the Royal Devon University Healthcare NHS Foundation Trust. The clinical team in Exeter has been reviewing all referrals from Somerset to manage this transition and ensure the service is not overwhelmed.

This letter summarises:

1. the referral criteria
2. how to refer to Clinical Genetics
3. other investigations to arrange alongside some referrals to cardiac genetics
4. some information on genetic testing where it is appropriate for testing to be arranged by non-genetics specialists

We recognise some of this information may be a change from your existing pathways. Please contact us if specific support is required either via email rduh.pcgreferrals@nhs.net or phone 01392 405745.

1. REFERRAL CRITERIA

Referrals are accepted in line with national guidance[1]. Whilst it is impossible to give an exhaustive list of which patients should be referred, the following is a guide:

Examples of appropriate referrals

- Patients wanting to discuss predictive genetic testing for a known genetic condition in their family
- Pregnancies with foetal abnormalities or where there is a family history of a genetic condition in a close relative
- Babies and children who are acutely unwell on NICU / PICU where there is a strong suspicion of a genetic condition or other acutely unwell children where R14 is being considered

- Patients in whom there is a strong clinical suspicion of a genetic condition but standard of care testing in line with the National Genomic Test Directory has failed to identify a diagnosis
- Where there is uncertainty regarding the diagnostic validity of the genomic variant and a clinical geneticist can help with genotype-phenotype correlation
- Patients with a likely pathogenic or pathogenic genomic variant requiring genetic counselling about risk to their relatives and/or reproductive options
- Patients with a likely pathogenic or pathogenic genomic variant causing a rare or complex multisystem disorder where the patient or their clinician would benefit from advice on management / genetic counselling
- Patients with a genetic disorder may benefit from referral when they transition between child and adult services, to discuss their condition in their own right and future reproductive options, where applicable

Examples of inappropriate referrals which will usually be declined:

- Straightforward diagnostic genetic testing for a specific gene or panel that can be requested by non-genetic clinicians (refer to section on genetic testing later on in this letter)
- Pharmacogenomic testing or interpretation of a pharmacogenomic test result in relation to prescribing specific drugs
- Common polygenic disorders such as isolated autistic spectrum disorder and autoimmune conditions
- Interpretation of a genetic test result from an unaccredited laboratory e.g. direct to consumer genomic tests, unless the test suggested a likely pathogenic or pathogenic variant for a condition we would normally test for e.g. a pathogenic BRCA variant
- Individuals with a somatic variant identified in the tumour where germline confirmation can be requested by non-genetic clinicians in the first instance or the somatic variant is not recognised to be associated with a germline predisposition to cancer
- Individuals at moderate risk of breast cancer in the absence of a confirmed familial heritable genetic risk factor - follow NICE guidance
- Patients better seen by another local specialty e.g. monogenic diabetes by Exeter expert diabetes team
- Some haematology conditions already managed by local haematologists
- Hypermobility type Ehlers Danlos syndrome without features suggesting an alternative diagnosis. See website for guidance.
- Familial Hypercholesterolaemia – refer to local lipid clinic
- Hereditary Haemochromatosis, unless atypical. GPs can arrange testing when indicated. See website for guidance.
- Alpha 1 antitrypsin deficiency, unless atypical. GPs can arrange testing where the diagnosis is suspected or in 1st degree relatives of affected individuals.
- Factor V Leiden and other thrombophilias – refer to haematology where indicated
- MTHFR (methylenetetrahydrofolate reductase) - NHS testing is unavailable
- Suspected foetal alcohol syndrome unless dysmorphic features are present and a clinical geneticist review is needed to review if the diagnosis is FAS or another condition

[1]Inclusion and exclusion criteria are regularly reviewed and updated as required and are currently based on the criteria set out in the document “Maximising the Patient Benefit of Genomics – the evolving role of the Clinical Genetics Services” published in December 2024

https://www.clingensoc.org/news/cgs-report-2024-maximising-the-patient-benefit-of-genomics-the-evolving-role-of-the-clinical-genetics-services/

2. HOW TO REFER TO CLINICAL GENETICS

Referrals from primary care should be sent via ERS

Non-primary care referrals should be sent by email to: rduh.pcgreferrals@nhs.net

Information to include in referrals

Please provide detailed clinical information in all referrals

- When referring following genetic testing please include the genetic report(s) with the referral
- When referring because of a family history please include full details of the family history
- For patients being referred because a relative has a specific genetic condition please include details of the diagnosis, full name and DOB of the affected relative(s) and which Clinical Genetics service saw them if known
- If the patient has been sent a 'dear relative' letter include a copy with the referral
- For cancer family history referrals please include details of all affected relatives including site and type of cancer and age at diagnosis
- Please include full name, DOB, address, telephone number, GP surgery and NHS number

Urgent referrals

Since delivering the service, the team have seen a high number of referrals coming through that are inappropriately marked as 'urgent'. This is causing a significant backlog which means it is taking longer than usual to process clinically urgent referrals.

We recognise that some patients have been waiting a long time due to issues accessing the service in the past. However, we need to please ask that you only mark referrals as urgent where it is clinically appropriate to do so.

Examples of referral scenarios to mark as urgent include:

- the patient is pregnant
- an acutely unwell child referred from paediatrics
- terminally ill and near the end of life

This will allow the team to manage the numbers of urgent referrals coming into the service and ensure they can prioritise those truly urgent patients.

Please be assured that whilst the service tries to review urgent referrals immediately, all referrals are reviewed, triaged and prioritised within a few days.

3. OTHER INVESTIGATIONS WHEN REFERRING TO CARDIAC GENETICS

- If referring a 1st degree relative of unexplained sudden death, please also do an ECG and refer to a cardiologist with expertise in Inherited Cardiac Conditions (ICC)
- If referring for assessment of possible Marfan syndrome please request an echocardiogram including aortic measurement at the sinus of Valsalva. If there are abnormalities please refer to cardiology also.

- If referring for predictive testing for an inherited cardiac condition, clinical cardiac screening is still also required. This should ideally be overseen by a cardiologist with an interest in inherited cardiac conditions. Please arrange this if your patient is not already receiving screening. If they are symptomatic please flag this with the cardiologist you refer to. Please send a copy of reports to Clinical Genetics. If you already have recent screening reports please enclose these with your referral.

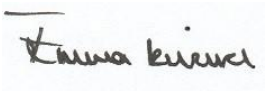
4. GENETIC TESTING

- Clinical Genetics does not see patients for straightforward diagnostic genetic testing for a specific gene, panel or whole genome sequencing (WGS) where the referrer is able to request the test (e.g. a neurologist requesting a genetic test relating to neurological disorder)
- For information on test eligibility and which specialties can request a test see Rare and inherited disease eligibility criteria in the National Genomic Test Directory: https://www.england.nhs.uk/publication/national-genomic-test-directories/
- Request forms can be downloaded from the laboratory website: https://www.nbt.nhs.uk/south-west-genomic-laboratory-hub/swglh-sample-test-information/swglh-sample-requirements-transport
- If advice is needed about which test to request please contact the laboratory: SWGLHenquiries@nbt.nhs.uk
- If advice or training is needed about how to request genetic testing, the genomic healthcare practitioners can often help (email rduh.swgenomicpractitioner@nhs.net)
- Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the patient and /or their family
- Individuals with disorders for which genetic testing is not recommended based on the National Genomic Test Directory should not be offered testing in England or elsewhere
- If a likely pathogenic / pathogenic variant is identified patients should be referred for genetic counselling, including for a discussion on risk to relatives and reproductive options. Patients with rare / complex conditions may benefit from a clinical geneticist review to discuss management and prognosis
- If the standard genetic testing is negative but there is a strong suspicion of a genetic condition discussion at an MDT and/or referral to Clinical Genetics may be appropriate
- A negative genetic test (even whole genome testing) cannot exclude a genetic condition

More information can be found on our website under ‘referral information for healthcare professionals’: royaldevon.nhs.uk/peninsula-clinical-genetics/

Thank you for your support. We look forward to working with you going forwards.

Yours faithfully,



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