

Genetic Referral - 2026 Draft

Introduction

This tool is in DRAFT and only to be used in Trial.

General Comments and Directions

- These criteria only apply to the triage of referrals to the Genetic Service.
- Scoring should be based on the considered view of the clinician taking into account the information provided in the referral, including the patient's history, examination, results of investigations, and the clinician's experience in treating like patients.
- The score should be calculated during the triage process and the patient and the referring clinician advised of the outcome.
- Where information is not provided and is not "known", or cannot reasonably be inferred, the lowest category should be assigned.

Clinical Override

- Clinical override may be appropriate in some circumstances if:
 - In the clinician's judgment there are exceptional factors not included in the prioritisation criteria, that the booking status generated does not adequately reflect the patient's priority, or
 - The patient's condition is an important example required for training and development of registrars or junior staff.
- The reason for the exception must be documented.
- It is important that Clinical Override is used judiciously.

Special cases

Decline with letter

In the following cases, the referral should be declined with a letter to the referring clinician.

- Referral for individuals with a sporadic chromosome rearrangement detected in products of conception
- Referral for diagnostic testing is able to be performed by referring service or has been mainstreamed

Automatic priority

In the case of either pregnant women, or where the referral is for a post mortem foetal assessment, the foetus and mother are considered as a single individual.

- Referral of patient where foetal post-mortem examination suggests a genetic cause
- Referral of pregnant woman with foetal anomalies where there is a suspicion of a genetic cause (acute)
- Referral for discussion of prenatal diagnosis in a current pregnancy (acute)
- Referral of patient with high grade serous ovarian cancer and eligible for PARP inhibitors

- Not a special case

Prioritisation

Genetic Referral

Where information is not provided and cannot be inferred, enter as lowest category.

Current status of referred individual

If the diagnosis cannot be assumed use “Moderate impact”. May consider psychological impact if stated in referral.

- No intellectual or physical impact (i.e. minor symptoms, little to no functional impairment, normal to mild cognitive challenges)
- Some intellectual or physical impact (i.e. noticeable physical difficulties, may require some medical support, learning disabilities, mild-moderate intellectual disability)
- Significant intellectual or physical impact (i.e. major or life-threatening physical impairments, highly dependant on medical care. Severe intellectual or physical disability with little to no independence.)

Likelihood of adverse outcome in 6 months

“Adverse health outcome” refers to deterioration of current condition or appearance of a new symptom associated with genetic disorder.

- Low (less than 10% – i.e. PT for most cancer predisposition syndromes)
- Moderate (between 10% and 50%)
- High (greater than 50%)

Impact of adverse health outcome

“Adverse health outcome” refers to deterioration of current condition or appearance of a new symptom associated with genetic disorder.

- No/mild impact (i.e. minor symptoms, little to no functional impairment, normal to mild cognitive challenges)
- Moderate impact (i.e. noticeable physical difficulties, may require some medical support, learning disabilities, mild-moderate intellectual disability)
- Severe/profound impact (i.e. noticeable physical difficulties, may require some medical support, learning disabilities, mild-moderate intellectual disability)

Potential future status of family members

If the diagnosis cannot be assumed use “Moderate impact”. May consider psychological impact if stated in referral.

- No intellectual or physical impact expected (i.e. minor symptoms, little to no functional impairment, normal to mild cognitive challenges)
- Some intellectual or physical impact expected (i.e. noticeable physical difficulties, may require some medical support, learning disabilities, mild-moderate intellectual disability)
- Significant intellectual or physical impact expected (i.e. major or life-threatening physical impairments, highly dependant on medical care; severe intellectual or physical disability with little to no independence)

Impact on management (for individual)

- Genetic information (unlikely to change management)
- May provide future benefit (e.g. improved surveillance/screening, PGT)
- May provide immediate benefit (extent of surgery)
- May allow targeted treatment (gene/variant specific treatments i.e. gene therapy/targeted treatments)

Information benefit for patient and/or family

It is presumed that basic reproductive advice is provided when appropriate in the general genetic advice. Specific reproductive information is information on the appropriateness, risks and benefits of PGT for example.

- Assessment unlikely to provide information benefit
- Increased knowledge and understanding only
- And, may inform life choices (non-reproductive)
- And, involves specific reproductive decision making (only choose this option where reproductive decision making is mentioned as a reason for referral, or when implied by referral from e.g. obstetrician or Fertility specialists.)

Expected time frame of changed management (for individual)

Include reproductive decision making as a change in management where mentioned as a reason for referral, or when implied by referral from e.g. obstetrician or Fertility specialists

- No Impact in management (genetic information only)
- Change in management 10+ years away
- Change in management 5-10 years away
- Change in management 2-5 years away
- Changed management appropriate soon (< 2 years)
- Immediate decision on management