Reasons for rejection of referrals from Primary Care to Clinical Genetics

- Referrals for a family history of cancer where the risk is not high. We will re-direct to a family history clinic for enhanced screening or give advice and guidance
- Common genetic conditions where testing can be done in primary care
  - Haemochromatosis
  - Alpha 1 antitrypsin
- Referrals for genetic conditions where testing is always done in secondary care
  - Haematological conditions e.g. sickle cell, thalassaemia, haemophilia, factor V Leiden
  - Familial hypercholesterolaemia

Reasons for rejection of referrals from Primary Care to Clinical Genetics

- Hypermobility or Type 3 Ehlers-Danlos syndrome
  - All the evidence suggests that this is not a single gene disorder and there is no diagnostic genetic testing for it as no causative genes have ever been identified
  - Additional features that would be concerning include:
    - Learning difficulties
    - Dysmorphic features
    - Pectus deformity, scoliosis or kyphoscoliosis
    - Cardiovascular features including aortic dilatation, arterial aneurysms or dissections, valvular abnormalities

Reasons for rejection of referrals from Primary Care to Clinical Genetics

- Diagnostic testing in unaffected individuals where no pathogenic variant has been identified in a family member. Diagnostic testing can never be used as an exclusion test
- Testing for susceptibility genes e.g. HLA haplotypes. These do not give any prognostic information. They may be used in specialities such as rheumatology to inform diagnosis and direct treatment in affected individuals
- Results of Direct to Consumer testing