Request title: Genetics Testing

Date of Response: 05th December 2016

Further to your Freedom of Information request, the Trust has answered your questions in the order they appear in your request.

Request

1. The number of patients diagnosed with genetic haemochromatosis who have been provided with genetic testing for haemaochromatosis and/or genetic counselling in relation to their condition.
2. The number of marriage or civil partners of patients diagnosed with genetic haemochromatosis who have been provided with genetic testing for haemochromatosis and/or genetic counselling in relation to their partner's condition
3. The number of siblings of patients diagnosed with genetic haemochromatosis who have been provided with genetic testing for haemochromatosis and/or genetic counselling in relation to their sibling's condition.
4. A copy of your protocol or pathway applicable to the provision of genetic testing/counselling for families of patients diagnosed with genetic haemochromatosis.

Reply

1. Thirty people were referred to the Peninsula Clinical Genetics service in relation to haemochromatosis during period 1 January 2015 to 31st December 2015. It is not possible to provide specific numbers of patients for request A,B and C due to the way data is captured on the patient management system.
2. It is not possible to provide specific numbers of patients for request A, B and C due to the way data is captured on the patient management system.
3. It is not possible to provide specific numbers of patients for request A, B and C due to the way data is captured on the patient management system.
4. The Exeter Molecular Genetics laboratory follows the attached 'EMQN best practice guidelines for the molecular genetic diagnosis of hereditary haemochromatosis (HH)'. It should be noted that, whilst this advises not to test partners, this may be undertaken by Clinical Genetics if there are several children and testing the partner would negate the need to test all children if the partner is negative.