Service information: **Idiopathic Congenital Nystagmus (NYS1)**

**Gene/Locus:** *FRMD7* Xq26.2  
**OMIM:** #310700

**Alternative Name:**  
- X-linked Congenital Nystagmus 1  
- Congenital Motor Nystagmus  
- Idiopathic Infantile Nystagmus

**Referrals:** Clinical Geneticists, Ophthalmologists

**Testing:** Diagnostic testing offered in clinically affected patients, carrier testing in females with a family history of *FRMD7* mutations (carrier testing referrals are accepted from Clinical Genetics only)

**Target Reporting Times:**
- Diagnostic: 40 working days  
  £400
- Carrier: 10 working days  
  From Clinical Genetics only  
  £100

Please contact the laboratory if urgent testing is required

**Test Details:**

- **Mutation screening for *FRMD7* mutations:** includes bidirectional Sanger sequencing of the coding region and the adjacent splice donor/acceptor sites of the *FRMD7* gene and MLPA (multiplex ligation-dependent probe amplification) analysis to detect exonic deletions/duplications of the *FRMD7* gene (MRC Holland P269 kit).
- **Testing for known mutations** in individuals with a family history of *FRMD7* mutations by Sanger sequencing or MLPA, as appropriate.

**Service Details:**

Combined data from the Leicester University research group has estimated that 64% of families with idiopathic congenital nystagmus have mutations in *FRMD7*. The current screening methodology will detect all of the published *FRMD7* pathogenic mutations. However, if a mutation is not detected by the testing procedure, a diagnosis of idiopathic congenital nystagmus cannot be excluded, as the patient may have a mutation in another gene or in a region of the gene not covered by the above analysis.

Males who have a mutation in *FRMD7* will be affected with idiopathic congenital nystagmus. Female carriers are at approximately 50% risk of being affected.

**Sample Requirements:**

- **EDTA** blood sample (1-4 ml), labelled with patient’s full name, date of birth and NHS number or genomic DNA (50µl at ≥50ng/µl).
- Samples should be accompanied by a fully completed referral card which should include the patient’s full name, date of birth and NHS number.
- Please also include details of the test, relevant clinical details and full details of the referring clinician and centre

**Consent:**

Please note that in submitting a sample, it is the responsibility of the clinician to ensure that consent has been taken i) for testing, ii) for storage, and iii) for the use of this sample and the information generated to be shared with the patient’s relatives and their health professionals. Following testing, a sample of the patient’s DNA may also be used anonymously to validate new tests and for internal quality control purposes.

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