

Date: 22nd October 2015

**RE: Changes to Testing Pathway for Fragile X Syndrome**

Dear Colleague,

The vast majority of referrals for the molecular testing of DNA from children with learning difficulties, intellectual disability and/or autism ask for BOTH Fragile X syndrome testing and a molecular karyotype (also known as microarray testing).

Recently, staff of Diagnostic Genetics (LabPLUS) conducted an audit covering a 4 year period of referrals for Fragile X testing. This revealed a detection rate of only 0.6% for *FMR1* CGG repeat expansions >200 (with no known family history) compared with a 5.4% detection rate of clinically significant abnormalities by microarray\*.

We wish to ensure that inappropriate testing is minimised to enable funds to be available to help develop other genetic tests that will be of benefit to families. In conjunction with the Genetic Health Service of New Zealand (Northern Hub), we have made a commitment to ensure that testing for Fragile X is undertaken appropriately.

**Hence, from 1<sup>st</sup> November 2015:**

- All routine requests for both microarray and Fragile X tests will be tested by **microarray in the first instance**.
- **Fragile X testing will only be performed if there is a known family history or if the clinical features meet the UKGTN Fragile X testing criteria.**
- The exclusion criteria may be helpful in deciding when **not to** test a patient.
- It is unlikely that there will be physical features suggestive of Fragile X syndrome in children under 5 years of age and for this category a positive family history of learning difficulties is a better indication for testing.

We would be grateful if you could bring this new policy and the indications for Fragile X testing to the attention of your teams, in particular to your junior colleagues. For completeness, we also attach the UKGTN approved testing criteria for Fragile X syndrome, which are **also available online at [www.ukgtn.nhs.uk](http://www.ukgtn.nhs.uk)**.

We trust this information will be helpful. If you have any queries please do not hesitate to contact the Director or Technical Head of Diagnostic Genetics, LabPLUS.

Yours sincerely,



Steve Absalom, Clinical Director  
LabPLUS



Dr Ian Hayes, Consultant Clinical Geneticist  
Genetic Health Service NZ Northern Hub

Cc:

Dr Don Love - Director, Diagnostic Genetics, LabPLUS, Auckland

Alice George - Technical Head, Diagnostic Genetics, LabPLUS, Auckland

Reference:

\*Developmental delay referrals and the roles of Fragile X testing and molecular karyotyping: A New Zealand perspective. *Elaine Doherty, Rachel O'Connor, Anna Zhang, Christina Lim, Jennifer M. Love, Fern Ashton, Karen Claxton, Nerine Gregersen, Alice M. George and Donald R. Love*. Molecular Medicine Reports 2013; 7(5): 1710-1714. DOI: 10.3892/mmr.2013.1386

Enclosed:

UKGTN Fragile X Mental Retardation Syndrome Testing Criteria Males

UKGTN Fragile X Mental Retardation Syndrome Testing Criteria Females

UKGTN Fragile X Mental Retardation Syndrome Testing Criteria Family History

Also available online at [www.ukgtn.nhs.uk](http://www.ukgtn.nhs.uk)