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Dear Colleague

Every year, we receive a large number of test requests for Fragile X syndrome and karyotyping in children with learning difficulties. The detection rate for a positive result on Fragile X testing is less than 1%. This low detection rate is partly a consequence of testing children who do not appear to comply with the nationally approved testing criteria for Fragile X.

Recently, we conducted an audit covering a 20 month period of referrals from our region where both Fragile X testing and array CGH testing was requested on the same sample. This revealed a detection rate of only 0.5% for Fragile X compared with a 4% detection rate of clinically significant abnormalities by aCGH.

We wish to ensure that inappropriate testing is minimised to enable funds to be released to help develop other genetic tests that will be of benefit to families.

In discussion with the four Regional Genetic Services in London and the Genetics Consortium, we have made a commitment to ensure that testing for Fragile X is undertaken appropriately. Hence, from **1st April 2012**:

- All routine requests for both aCGH and Fragile X will be tested by aCGH in the first instance
- Fragile X testing will only be undertaken **if the aCGH shows no pathogenic abnormality and the clinical features meet the 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.
- This policy is being implemented across all genetic services in London.

We would be grateful if you could bring this new policy, and the indications for Fragile X testing, to the attention of your teams and in particular to your junior colleagues. For completeness, I also attach the UKGTN approved testing criteria for Fragile X syndrome.

We trust this information will be helpful. If you have any queries please do not hesitate to contact us.

Page 2 of 2

Yours sincerely,

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Enc: UKGTN testing criteria

c.c.

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