Alberta Health Services

# Laboratory Bulletin

**Date:** **June 8, 2015**
**To:** Edmonton Medical Genetics Clinic
**From:** Genetic Laboratory Services - North, Molecular and Cytogenetic Laboratories
**Re:** Copy Number Variant (CNV) Follow-up Policy

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**Key Messages:**
This bulletin is to communicate an up-dated policy regarding the parental follow-up of genomic copy number variants (CNVs) by the Genetic Laboratory Services North Molecular and Cytogenetic Laboratories.

With respect to CNVs identified by microarray:

1. Follow-up of microarray CNVs will be performed using FISH, qPCR, MLPA or microarray as indicated based on the size and location of the CNV
2. Parental testing to determine if a CNV is de novo or familial will be performed using the same technology as the verification in the proband
3. Whenever possible, FISH analysis will be performed on the proband and the parents to provide structural information of clinically significant CNVs
4. When FISH testing is not possible, the small possibility of a structural chromosome rearrangement cannot be excluded.

This policy is consistent with CCMG Guidelines for Genomic Microarray Testing, 2010

With respect to deletions/duplication identified by targeted qPCR or MLPA methods (example: 22q11 deletions):

1. Parental testing, if indicated, will be performed using the same technology used to identify the deletion/duplication in the proband
2. If the testing for the proband and parents is done by qPCR or MLPA, it will not be possible to rule out the small possibility of a structural chromosome rearrangement
3. Prenatal testing for the CNV will be available by the same method as the proband. Prenatal testing is available regardless of parental status due to the 3-4% risk of gonadal mosaicism.

The laboratory uses a threshold of a 1% risk as a guideline principal. The likelihood of a structural rearrangement that will not be detected using the above algorithm is <1% based on the available literature. If the family or medical history is suggestive of an increased risk of a structural rearrangement, a karyotype may be indicated.

**Why this is important:**

- This information will allow clinicians to provide the appropriate counselling and testing options for their patients.

**Action Required:**

- If the family or medical history is suggestive of an increased risk of a structural chromosome rearrangement, please include the relevant information on the requisition form

**For additional questions contact:**

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**This bulletin has been reviewed and approved by:**

- Dr. Martin Somerville, Medical Scientific Director, Genetic Laboratory Services