HAMILTON REGIONAL LABORATORY MEDICINE PROGRAM

DATE: November 22, 2012

TO: Paediatricians with Hamilton Health Sciences & St. Joseph's Healthcare
Family Physicians with Hamilton Health Sciences & St. Joseph's Healthcare
Prenatal Services at Hamilton Health Sciences
Laboratory Reference Centre Clients

FROM: Dr. Elizabeth McCready, Head of Molecular Genetics

RE: **CHANGES TO CHROMOSOME MICROARRAY (CMA) TESTING**

Effective November 26,2012 the McMaster/HRLMP Clinical Genetics Laboratory will be changing the platform being used for chromosome microarray (CMA) testing from the BlueGnome CytoChip ISCA 4x180K platform to the Affymetrix Cytoscan HD platform.

Effective January 1, 2013, CMA testing will be performed for referrals related to developmental delay, intellectual disability, autism spectrum disorder, neurobehavioural difficulties, and multiple congenital anomalies. Chromosome studies will no longer be available for these indications except in cases requiring follow-up of previously identified familial chromosome rearrangements or if a specific chromosome trisomy syndrome is suspected. FISH will continue to be available for instances where a specific microdeletion is suspected.

Meta-analysis of publications examining the use of CMA testing for the clinical investigation of individuals with developmental delay, intellectual disability, autism spectrum disorder and/or multiple congenital anomalies has shown that CMA has an improved detection rate (12.2%) compared to classical cytogenetic techniques (3%). As such CMA testing is recommended as a first-tier test in the clinical investigation of individuals with these indications [Miller *et al*. (2010). *AJHG*, 86:749-764].

PLEASE NOTE: This change to the CMA test procedure allows for improved sensitivity and detection of a broader range of chromosome aberrations than previous versions of the CMA test or classical cytogenetic techniques, including detection of regions of homozygosity in the genome that may be associated with imprinting or recessive disorders due to either consanguinity or identity-by-descent. An option to opt-out from receiving the results of homozygosity testing will be available on the requisition form.

The new CMA platform requires collection of peripheral blood in EDTA tubes (rather than the sodium heparin tubes used for classical cytogenetic studies). Microarray requisitions will be available through the HRLMP website (http://www.hhsc.ca/body.cfm?id=239). **All requisitions for CMA testing submitted to the HRLMP Molecular Cytogenetic Laboratory should be accompanied by 5-10 cc of peripheral blood collected in an EDTA tube (or a minimum of 3 cc for infants and neonates under 1 year of age**). The accurate interpretation and reporting of CMA genetic test results are contingent upon awareness of the reason for referral and clinical information provided; to provide the best possible service health practitioners are encouraged to provide as complete clinical information on the requisition as possible.

Thank you for your understanding regarding this change of service. If you have any questions please do not hesitate to contact the laboratory or myself directly at 905-521-2100 ext 73706 Fax: 905-521-2651 mccready@hhsc.ca.