

NEWS RELEASE

For immediate release

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BC Children's and BC Women's hospitals aim to improve the care for children with congenital intellectual disabilities

*Research team looking for new chromosomal indicators to aid early identification of the
cause of intellectual disability*

Vancouver, B.C. – Researchers at BC Children's Hospital and BC Women's Hospital, UBC and Queens University in Ontario have received \$584,000 in funding from the Canadian Institutes of Health Research to study children born with an intellectual disability (ID) of unknown cause.

"Our study hopes to link previously unknown genetic causes of ID to specific clinical characteristics," said Dr. Evica Rajcan-Separovic, a cytogeneticist with the Children's Department of Pathology and researcher with the BC Research Institute for Children's and Women's Health (BCRICWH). "Making those links will allow us to create new health watch guidelines for each previously unknown cause of congenital ID that we identify. Those new guidelines would be used to help doctors diagnose children who are not meeting developmental milestones."

Dr. Suzanne Lewis, a medical geneticist with the Women's Department of Medical Genetics, researcher with the BCRICWH and a co-investigator in the study, hopes that improving the overall knowledge and clinical practice related to each new condition will improve the quality of life for those with a congenital ID. "Knowing in advance how a person's abilities and/or health may be affected makes it a lot easier for families and physicians to care for these children, and help them reach their full potential. It also gives parents an answer regarding what's going on with their child and hopefully reduces the number of tests those children may undergo."

ID is the diagnosis given to persons who have significant cognitive and adaptive impairment that starts early in life. Congenital IDs affect one to three per cent of the Canadian population. The causes for some IDs are known (Down syndrome, fragile X), but causes for the remaining 30 to 50 per cent remain unknown.

The research team led by Dr. Rajcan-Separovic, with Dr. Suzanne Lewis and Dr. Jeanette Holden (Queens University) as co-investigators, will look for sub-microscopic deletions and duplications on the chromosomes of children with ID utilizing the newly-developed technology of genomic microarrays.

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Genomic microarrays are very short segments of DNA from all 46 human chromosomes, arrayed on glass slides. The arrays used in the study will provide a resolution 10-20 times higher than routine chromosome analysis and allow identification of previously “invisible” chromosomal changes at the whole genome level.

An earlier pilot study confirmed that this approach will identify submicroscopic abnormalities in 15-20% of individuals with ID of unknown cause. The study, to be performed at BC Children’s, will recruit patients up to 18 years of age with ID of unknown cause and screen their genetic material for previously undetected sub-microscopic chromosomal changes.

An additional 1,000 subjects with ID and 100 control subjects will then be screened for the newly detected sub-microscopic chromosomal changes to identify other individuals with the same findings and clinical features. These results will further assist the research team to better recognize and understand the clinical characteristics and related anticipatory medical issues attributable to each newly-identified genetic variation, as well as how frequently each finding appears in the ID and general populations.

BC Children’s Hospital, an agency of the Provincial Health Services Authority, provides expert care for the province’s most seriously ill or injured children, including newborns and adolescents. For more information please visit www.cw.bc.ca.

BC Women’s Hospital & Health Centre, an agency of the Provincial Health Services Authority, provides service for pregnant women, newborns and women with specialized health needs from across the province. For more information please visit www.cw.bc.ca.

BC Research Institute for Children’s and Women’s Health operates in partnership with UBC and the Children’s & Women’s Health Centre of British Columbia, an agency of the Provincial Health Services Authority.

The Provincial Health Services Authority (PHSA) plans, manages and evaluates selected specialty and province-wide health care services across BC, working with the five geographic health authorities to deliver province-wide solutions that improve the health of British Columbians. For more information please visit www.phsa.ca.

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Note to editors: Dr. Evica Rajcan-Separovic is available for interviews.

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