

## **Novel Technology for Fragile X Population Screening**

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Mutations in *FMR1* gene cause Fragile X syndrome (FXS) which is a common heritable cause of intellectual disability, and is often associated with autism. Early treatment and/or behavioral intervention as the result of diagnosis of FXS, which would also allow for identification of genetic risk for subsequent pregnancies and other relatives are strong arguments in favor of newborn population screening for this syndrome. As the degree of intellectual disability and behavioral problems in females with *FMR1* mutations is highly variable, there is also a need for a simple test that can be used to identify those affected and thus most likely to benefit from early intervention. Currently there is no molecular test that reflects behavioral / intellectual deficits in FXS girls and boys with and without autism. The work presented at the Fragile X Research Symposium describes a novel high-throughput FXS test which has been developed at the Victorian Clinical Genetics Services, Murdoch Childrens Research Institute, in collaboration with School of Psychological Science, La Trobe University. This test has a great potential to be used for early detection of FXS in either males or females, with clear implication for inclusion of this syndrome into newborn population screening program. Another potential application of this test is in targeted FXS diagnostics in young children. Preliminary results also show that the test works in non-invasive saliva format as well as in blood. Since obtaining a blood sample, particularly in young children with intellectual disability and autism can be difficult for the clinician and traumatic for the child, if confirmed in our ongoing studies, the test can also offer a maximally non-invasive option making it more attractive for improved targeted FXS diagnostics.