Copy number variations (CNV) has extensively been shown to cause pathogenic disorders. The project we are running have focused on developmental delay caused by submicroscopic CNV in the human genome. It is based on an epidemiologic collection of patients (n=137) with severe mental retardation (IQ <50) derived from the southwest of Sweden (county of Halland) between the years 2003-2007. In 21 children with prenatal aetiology, specific diagnosis was missing, and these children will therefore be analysed by high resolution SNP array analysis for CNVs. This study aims to make an epidemiologic report for children with SMR and may indicate improvements of the clinical workflow for these children. Furthermore we will evaluate the usefulness of array analyses for CNVs.

**Publikationer i vetenskapliga tidskrifter**


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