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Interpretation of clinical relevance of X-chromosome copy number variations identified in a large cohort of individuals with cognitive disorders and/or congenital anomalies

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Abstract:
Genome-wide array studies are now routinely being used in the evaluation of patients with cognitive disorders (CD) and/or congenital anomalies (CA). Therefore, inevitably each clinician is confronted with the challenging task of the interpretation of copy number variations detected by genome-wide array platforms in a diagnostic setting. Clinical interpretation of autosomal copy number variations is already challenging, but assessment of the clinical relevance of copy number variations of the X-chromosome is even more complex. This study provides an overview of the X-Chromosome copy number variations that we have identified by genome-wide array analysis in a large cohort of 4407 male and female patients.We have made an interpretation of the clinical relevance of each of these copy number variations based on well-defined criteria and previous reports in literature and databases. The prevalence of X-chromosome copy number variations in this cohort was 57/4407 (w1.3%), of which 15 (0.3%) were interpreted as (likely) pathogenic.