# P-711 - GENETIC SYNDROMES AND THE AUTISM SPECTRUM

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**Introduction:** Autistic features are present in a variety of genetic syndromes and have stimulated research into the genetic underpinnings of autism spectrum disorders. To understand this genetic heterogeneity, various techniques can be used such as cytogenetic research, linkage studies, and association studies. At present, CGH-microarray technique is primarily used, which enables examination of the complete genome for the presence of microdeletions and duplications or so-called copy-number variations (CNVs). The most investigated syndromes are fragile-X syndrome, Rett's disorder, tuberous sclerosis and 22q11microdeletion syndrome. In addition, metabolic disorders as well as a variety of somatic diseases may be accompanied by autistic symptoms.
**Objectives:** To study the genetics involved in autism spectrum disorders.
**Aims:** Review of the literature about the most investigated genetic syndromes.
**Method:** Data from own studies in patients with 22q11 microdeletion syndrome are briefly discussed against the background of the results from the literature on other genetic syndromes that are rather frequently related to autism.
**Results:** Fragile-X syndrome, Rett's disorder, tuberous sclerosis complex, 22q11 microdeletion syndrome, and metabolic disorders like X-linked creatine transporter deficiency syndrome, all deal with mutations or copy number variants in genes that code for proteins responsible for neuronal growth and maturation, and/or synaptic functioning.
**Conclusions:** From the vast amount of information, it becomes obvious that autism is a very heterogeneous disorder both phenotypically and genotypically, reason why the search for single high impact genes will not be very successful. Focus should be given to the endophenotype strategy to unravel potential gene-brain-behaviour relationships in autism spectrum disorders.



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