

This bachelor thesis deals with the analysis of clinical features in patients with autism spectrum disorders who were investigated using DNA microarrays. The introductory section is focused on the definition of autism and its subtypes, on currently known genetic causes of this neurodevelopmental disorder and on the possibilities of the laboratory diagnosis. Autism is likely caused by CNV occurring in different loci of the human genome, which can be efficiently diagnosed using DNA microarrays. This technique enables the detection of many CNV, but in most cases only common population polymorphisms can be identified. Our group consisted of 98 patients who suffered from some subtype of autism spectrum disorder. All patients were investigated using the microarray HumanCytoSNP-12 manufactured by Illumina. A retrospective analysis of clinical features of interest that were found in the medical documentation of the patients was performed. Statistical analysis of the data was performed to find possible associations. Specific pairs of features were compared in more detail. Features with known correlation previously published in the literature or features where a correlation could be expected were selected for this detailed analysis. Some findings were concordant with the published data, but some were not. Finally, it was determined whether there is a significant difference between the group of patients in whom an aberration associated with autism was identified and the group with no clinically relevant aberration. No significant association was found. Three patients are described in detail. These patients carried a causal aberration – a microdeletion/microduplication, which was described previously in several autistic patients. Namely it was a 15q11-q13 duplication, a deletion of the NSD1 gene and a MECP2 duplication.