

# Autism – aspects of the aetiology

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## ABSTRACT

The eight studies for this doctoral dissertation were carried out at Centre for Psychiatric Research, University of Aarhus. This thesis focused on the aetiology of autism spectrum disorders (ASDs) from an epidemiologic (register-based) and genetic point of view, and the purpose was to suggest and identify aetiological factors involved in ASD. ASDs are largely genetic in origin, with a complex mode of inheritance.

The occurrence of childhood autism and other ASDs was studied, and we found that the prevalence and incidence rates of ASDs have increased during the 1990s which most likely is due to changes in the registration procedures and increased awareness of the disorders.

We investigated whether individuals with autism more commonly than expected had chromosome abnormalities or medical conditions in order to localise chromosome regions of interest regarding the aetiology of autism. We found similar rates of medical disorders in autistic individuals as have been found in other studies and a significantly increased rate of congenital malformations. We also found regions on the autosomes which could contain genes involved in ASD (candidate regions).

In a study of risk factors, we found a significantly increased risk of autism in individuals with a history of autism or other ASDs in siblings, supporting the genetic component involved in the aetiology of autism. Significantly increased risk of autism was found if the mother had been diagnosed with a psychiatric disorder. Increased risk was also found if the mother was born outside Europe and if the parents were born in different countries. The risk of autism was associated with the child's place of birth in the capital or capital suburb and with increasing paternal, but not maternal, age. These variables may be by-proxy variables for factors important for the development of ASD.

In addition, two candidate genes were studied and no association with childhood autism could be detected. Furthermore, as one of the first autism research groups we used an isolated population on the Faroe Islands in a genome-wide scan to search for candidate regions for autism, and we identified some potential candidate regions for autism which need to be studied further in other populations.

In conclusion, we have contributed to the growing knowledge about autism aetiology, although at present only very few specific aetiological factors have yet been identified.