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Autism or autisms?

EDITORIAL

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The cause of autism is biological, but the diagnosis is made on the basis of observed behaviour. Autism populations are highly heterogeneous.

Autism is characterised by difficulties with communication and social interaction as well as repetitive and stereotypical behaviours. Medical treatment has no direct effect on the core symptoms, and autism is primarily treated through education and behavioural services. Neither biological tests nor blood tests can confirm the diagnosis, which is made on the basis of a thorough clinical examination and identifying the patient's behavioural profile. Early identification provides an opportunity for early treatment, but it is mainly children with delayed language and cognitive development who show clear symptoms of autism spectrum disorder (ASD) at an early stage. It is not possible to identify all children with autism before they reach school age (1).

In this edition of the Journal of the Norwegian Medical Association, Eig et al. have published a study conducted in the former county of Sør-Trøndelag on the prevalence of ASD among preschool children (2). The study found that the risk of being diagnosed with autism was seven times higher in children of mothers with non-Norwegian backgrounds compared to children of Norwegian-born mothers. It is not known why this is the case, but a Norwegian Official Report also documented an increased risk of autism diagnoses in many minority groups (1).

Some people with autism have average or above average intelligence, often referred to as Asperger syndrome, whereas others have a severe intellectual disability. The term *autism spectrum disorder* reflects the large variation in the

level of functioning. However, the terms 'autism' and 'autism spectrum disorder' are often used interchangeably. The heterogeneity in autism populations is mainly due to the fact that everyone's biological make-up is different. Moreover, when researching the causes of autism, it can be useful to study autism with and without intellectual disability as two separate groups. This applies when studying genetics as well as the association between immigration and autism risk, as Eig et al. have done (2).

An autism twin study published in 1977 was a breakthrough for the genetic recognition of autism (3). In some people, autism is due to one rare genetic variant with a high penetrance. Such genetic variants are found primarily in the group with intellectual disability. In the group with average or above average intelligence, the genetic predisposition for autism tends to be due to the sum of the effects of several common genetic variants, and each individual variant has little effect (i.e. a low penetrance). Sometimes the genetic predisposition can be explained by an interaction between rare and common variants (4).

> ***«It is important to note the difference between genetically defined diagnoses and behavioural diagnoses»***

Some rare genetic syndromes that are associated with intellectual disability are also associated with a high prevalence of autism (5). For example, 50 - 60 % of boys with fragile X syndrome meet the diagnostic criteria for autism (6), but this also means that there are many who do not. It is important to note the difference between genetically defined diagnoses and behavioural diagnoses. The large variation in genetically proven causes is why some experts talk about autisms rather than autism. Using the plural form of the word 'autism' emphasises the fact that individuals with autism are highly different from each other.

Some babies are born with a predisposition to develop autism. Genetic predisposition is the major contributing cause, but environmental factors can also play a role, particularly factors that have an impact before the baby is born. For example, it has been reported that obesity in mothers and use of valproate medication during pregnancy increase the risk, while folate supplements reduce the risk (4). Some environmental factors seem to be closely linked to genetics. Increased risk of autism associated with advanced parental age in mothers (> 40 years) and fathers (> 50 years) is believed to be due to the higher number of new genetic mutations in the gametes with advanced age (7).

Parents with a migrant background thus seem to be a risk factor for autism. This can be more difficult to explain biologically, and more research is needed in this field. The study by Eig et al. is relatively small, but international studies also report an association between migration and ASD (8). However, the increased risk only seems to apply when there is co-occurring intellectual disability. Several studies have reported a *negative* association between migration and the risk of autism without intellectual disability (8).

Language and cultural differences can make it difficult to detect and diagnose autism, and it is unknown whether all children with autism and average intellectual abilities are actually identified. Norwegian clinicians need to be

aware that some children with an immigrant background can be extra vulnerable. It is important that these children are identified and given the best possible help. We must also avoid stigmatising individual population groups.

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