Autism

Autism is a psychiatric disorder caused by abnormal development of the central nervous system . It is manifested by the inability to create social ties, communicate and complete communication with facial expressions, limited, stereotyped interests and inadequate response to sensory stimuli. Autism is most often diagnosed at the age of 3.

The term *autism* includes a clinically heterogeneous spectrum of diseases, which is referred to as **autism spectrum disorders (ASD)**. In the DSM-IV classification, ASD is divided into **Asperger syndrome**, **autistic disorder and PDD-NOS**. In the international classification of diseases, ICD-10, used in the Czech Republic, autism is classified under *pervasive developmental disorders* (F84) and includes **childhood autism** (F84.0) and **atypical autism** (F84.1). Asperger's syndrome is not directly characterized as autism in the ICD-10, being distinguished from autism by the absence of speech and cognitive deficits.

Symptomatology

The main three symptoms:

- **Qualitative disorders in reciprocal, social interaction** - Inability to "read" others, ignoring them. Avoiding eye contact, not trying to please others, or not sharing interests with them (e.g. bringing toys to parents, etc.). At home, they prefer solitude to company.
- **Impaired communication** – Reciprocal communication with speech, gestures, facial expressions does not develop. Young autistic children are unable to use gaze to communicate and attract parental attention. Inability to grasp the concept of a thing as a concept, too concrete understanding of concepts, inability to abstract ("teddy bear" is always a concept for one particular toy).
- **Narrow spectrum of interests** – Stereotypical activities (e.g. constant, tedious building of a tower of blocks), lack of interest in toys, and on the contrary, often an interest in technical objects (screwdriver, switch). Fantasy-free games without interaction with other children. Resistance to any changes, etc.

other symptoms

- **Hyper- or hyposensitivity** to sounds and touches (e.g. the sound of a vacuum cleaner can cause extreme discomfort, whereas some very painful stimuli can be ignored)
- **Peculiar food habits**, such as only eating French fries from McDonald's
- Abnormal sleep patterns
- Self-injurious tendencies in behavior
- Impaired motor development
- Failure to assess danger
- 50-70% of autistic children are identified as retarded on non-verbal IQ tests
- 25% develop *epilepsy*

Etiology and pathogenesis

Genetic background[1]

Autism is a polygenic disease involving genes on chromosomes 1, 2, 4, 7, 13, 15 and 16, e.g. FOXP2 (a transcription factor expressed in the developing and adult brain, as well as in the lungs and intestines, is key to the development of speech and language regions in embryogenesis), RELN (neuron migration in embryogenesis), HOXA1 (homebox gene, important for organization in the anteroposterior axis of the rhombencephala) and genes for GABA receptor subunits (GABRB3, GABRA5, GABRG3)[2], and others.

Enviromental influence

The effect of alcohol, thalidomide, and exposure to rubella virus on prenatal development before 30 weeks of gestation is discussed. The earlier "cold mother" hypothesis was *not confirmed*.

Disrupted development of the cerebellum and temporofrontal pathways

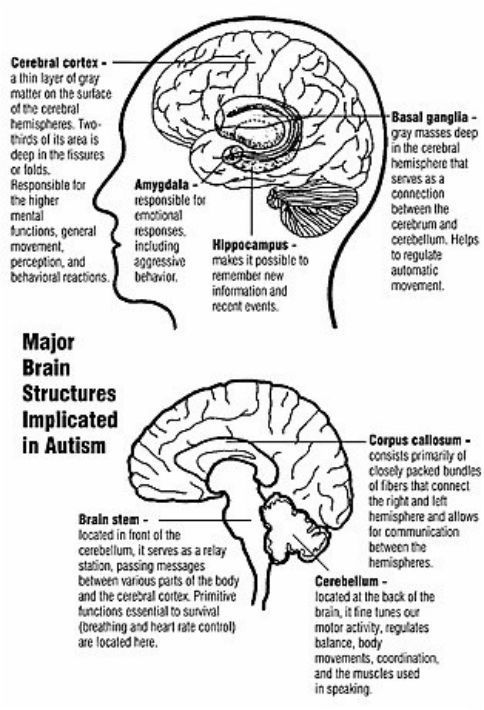
The cerebellum, frontal and temporal cortex, basal ganglia, and others appear to be important in the pathogenesis of autism. Some studies point to the fact that research that has been moving towards individual brain structures has so far yielded rather ambiguous results, and it is therefore better to look at autism as a connectivity problem.

Disrupted brain circuits[1]

Three fundamental brain circuits whose dysfunction can lead to autism are considered. On the one hand, it is the so-called **WCC (weak central coherence model)**, in which there is a discrepancy between local and global data processing. Autistic people are unable to use *the context of a situation* to help them better understand the environment.

In the second case, it is about *the Theory of mind*, which is key in the so-called mentalization, i.e. estimating the thought processes of other people, which enables the integration of the individual into society. Social cognition is impaired in autistic people, so they cannot read the emotions of others or their own.

A third system is the mirror neuron system, which is activated when a test subject observes a particular activity. **Mirror neurons** are apparently necessary for *non-verbal communication*, its encoding and decoding. It is not without interest that the cerebellum and frontal cortex play their role in all three mentioned systems.



Areas of the brain whose dysfunction can lead to autism

Role of the cerebellum[3]

The involvement of the cerebellum in individual circuits is discussed above. Attention is focused here on the cerebellar cortex, especially the Purkinje cells. **Magnetic resonance spectroscopy** revealed a decrease in *N-Acetyl-Aspartate* in the cerebellar cortex, which corresponds to a reduction in the number of Purkinje cells found in cross-sectional studies. Decreased levels of *Bcl-2* (inhibitor of apoptosis), *Reelin protein* (coded by the RELN gene, see above), *NCAM* (neural cell adhesion molecule) and *NT3* were found , while the levels of VIP , NT4/5, BDNF and CGRP were increased. A **reduced density** of Purkinje cells is apparently key to the further development of autism. The most frequently affected parts of the cerebellum are the vermin tuber and pyramid (VI and VII), on the other hand, involvement of lobules IV is rare, which is in accordance with the different development of these parts of the cerebellum. Impairment of the tuber and pyramid is more evident in low-functioning autism, while it may not be expressed in high-functioning autism. Despite all this, however, IQ and cerebellum morphology cannot be clearly related to each other, even though the cerebellum plays one of the key roles in the pathophysiology of autism. A *smaller cerebellar volume* is inversely related to the size of the frontal lobe, from recent research it seems that cerebellar dysfunction precedes the involvement of the frontal lobes.

Frontal lobe

Certain areas of the frontal lobes have been linked to autism. Activity in the pars opercularis is inversely proportional to the severity of the disability. It can be judged that a disturbed system of mirror neurons is one of the causes of autistic "trapping in one's own world." Connections with (expressive) speech functions are reported. Other frontal lobe areas considered in autism: gyrus frontalis inferior and others.

Other structures

Temporal lobe, amygdala, basal ganglia, insula, gyrus fusiformis, hippocampus, etc.[4]

Links

Related Articles

External links

- Autismus (czech wikipedia)
- Autism (english wikipedia)

- Autism Spectrum Disorders (https://www.ncbi.nlm.nih.gov/books/NBK1442/)
- GeneCards – FOXP2 (https://www.genecards.org/cgi-bin/carddisp.pl?gene=FOXP2&search=FOXP2)
- GeneCards – RELN (https://www.genecards.org/cgi-bin/carddisp.pl?gene=RELN&search=RELN)

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