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Review of anatomical and histological alterations in the brain of patients with autism

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Abstract

Autism spectrum disorder is a developmental illness which interferes with communication and behavior. autism can be detected at any age but symptoms become clinically evident in the first 2 years of life. The main symptoms are associated with social communication, interaction, monotonus actions and loss of enjoyment in objects and events. In addition it is linked to other psychological disorder like attention-deficit hyperactivity and epilepsy in the early years of life. Early diagnosis of the disorder with successive interference is very helpful to refine verbal outcomes and upgrade critical symptoms. Nowadays it is evident the neuroanatomical involvement in this disorder in early childhood and teenage groups like the cortical tissue of frontal, temporal and parietal lobes, amygdala and hippocampus, in the inferior cerebellar vermis and left cerebellar lobule.

In conclusion: Autism is a neurodevelopmental disorder which characterized by diminished communication skills, behavioral disturbances and cognitive impairment issues which definitely associated with neuroanatomical and microstructural changes in the brain cortex. In addition to genetic role in its etiology.

Keywords: Autism; Brain; Neuroanatomy; Histology; Abnormalities

1. Introduction

Autism is now one of the most public neurodevelopmental disorders affecting children in the first 4 years of life (1). It characterized by late and interrupted and diminished communication skills, monotonous, repetitive and stereotype form of behavior, affection, attentiveness (2). It affects male about four times higher than female (3)

Early diagnosis of the disorder with successive interference is very helpful to refine verbal outcomes and upgrade critical symptoms (4) as the neuropsychological progress is greatly associated with the environment (5). The beginning and progression of the disorder is gradual and characterized by retardation in the developmental growing and basic communication capabilities (6).

In addition, parents as well need support because having autistic boy is very stressful and it may associate with parental depression (7). In some cases, parents may need to be enrolled in training courses to learn how to deal with their child because autism can create inappropriate communication with the autistic child (8).

It may associate with other psychological disorder such as attention-deficit hyperactivity disorder in which they share some neural and communication deficits with hyperkinesia (9). It also associated with epilepsy especially generalized seizure at early child hood and in teenage groups (10).

This review aims to determine the neuroanatomical and microstructural changes and genetic mutation role in the brain of autistic patients.

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2. The Neuroanatomical and Histological Abnormalities in Autism

The brain of autistic child revealed some anatomical abnormalities which are not found in other normal child (11). Many regions especially those associated with social communication and personal wellbeing were involved like the cortical tissue of frontal, temporal and parietal lobes, amygdala and hippocampus, in the inferior cerebellar vermis and left cerebellar lobule (12).

Courchesne et al. 1988 (13) found that there is severe decrease in the size and volume of cerebellar vermis with associated reduced cellular number, they suggested that this abnormality has been occurred during the brain developmental process which suggest genetic etiology of the disease.

It is found that the size of hippocampus is larger in autistic patients than in normal children while the thickness of amygdala and some parts of cerebellum is less than in normal children (14). Even the white matter microstructure is affected with abnormal tracts and in some cases volumetric decrease in corpus callosum size was also reported and this is directly related to abnormal connectivity between the two hemispheres of brain (15,16).

Some regions in the brain cortex showed rapid enlargement with enlarged brain volume so, there will be early hyper expansion with subsequent unusual decelerated growth (17)

Many studies on autistic children brains revealed decrease in the amount of gray matter in the basal forebrain especially in putamen, lobules of cerebellum, and insula which are very important in cognition and behavior. In addition to irregular connection of gray matter in some regions of the brain to basal forebrain (18).

Perry 2001 et al (19) found that M_1 receptor binding is 30% less in autism especially in parietal region. In addition, nicotine receptors are extremely reduced especially in frontal and parietal gray matter. Reduced number of Purkinje fiber layer cells is reported to result in autism like behavior in mice and aberrant fibers are also reported (20).

Amygdala of temporal lobe bilaterally are responsible for fear, angry, happiness, anxiety, emotional memory are found to enlarge in volume especially in the first few years of life later, in adulthood there will be decrease in its volume, with decrease the whole nerve cellular body size and appeared highly dense cells (21). The frontal cortex is responsible for mental, intellectual functions, social and emotional wellbeing and even education and learning abilities in the brain (22). Atypical growth forms of frontal cortex with unequal cortical thickening with neuronal cellular disruption with abnormal connection in the prefrontal region (23)

Carper and Courchesne (2000) (24) found that the impairment in cognition process in autistic patients is associated with neuroanatomical aberration in frontal lobe and or cerebellum.

Bauman and Kemper (2005) (25) found that many regions are affected in autistic brain especially the limbic system and hippocampus in which reduced neuronal cellular volume and highly dense cells were also detected, while in cerebellar cortex there was decrease in the percentage of Purkinje neurons and other cerebellar nuclei were also affected. Light-stained neuronal cells were detected especially in adulthood autistic patients.

Zhou et al (26) reported that the defect visual perception in autistic children is associated with incomplete developmental issues in visual area.

Several studies revealed the autism-linked genetic mutations and Many genes are detected like deletion in (DIA-1) gene. In addition, UBE3A gene is highly expressed on specific chromosome 15q11–q13 this may induce several gene replication and it may accompanied by gene deletion (27). *3709DelG gene mutation in the prefrontal region and it is related to increase in the head circumference (28)*.

Other genes reported in autism, rs1858830 gene polymorphism is associated with hyperactivity in limbic system specifically amygdala and striatum with white mater corruption (29). RELN gene is accompanied by severe lacking of specific amino acids in cerebellar cortex (30).

Stoner et al. (2014) (31) studied different cortical tissue in the brain and they found that some regions showed irregular gene expression *Abnormal* proliferation, differentiation, and relocation of nerve cells in an incorrect layer.

Rapanelli et al. (2023) (32) found that excision of Cul3 acetyl cholinergic nerve body cells in mice is greatly associated with cognition loss and reduced actions of these neurons in the basal forebrain and mutation in this gene is considered as a precipitating factor for autism.

In addition, in postmortem autistic patients, atypical gene expression in mRNA genes were detected (33).

3. Conclusion

Autism is a neurodevelopmental disorder which characterized by diminished communication skills, behavioral disturbances and cognitive impairment issues which definitely associated with neuroanatomical and microstructural changes in the brain cortex. In addition to genetic role in its etiology.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

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