

Generalized Overgrowth in Autism

Dr Mohammed Arif *PG candidate for MD pediatrics,*
Rajib Chatterjee, *Professor and HOU, PG guide, MBBS, DCH, MD pediatrics*
Dr Himanshu S raichandani *PG candidate for MD Pediatrics*
Department of Pediatrics Rural Medical College, PIMS (DU), LONI, Maharashtra

Date of Submission: 25-07-2022

Date of Acceptance: 08-08-2022

I. Introduction

Autism is a complex developmental disorder characterized by marked impairments in social interaction and communication accompanied by arrange of repetitive behaviors and restricted interests. Along with pervasive developmental disorder—not other wise specified (PDD-NOS), which is typically characterized by less severe impairments and more atypical presentation, autism belongs to the class known as autism spectrum disorders (ASDs). Although the diagnosis of ASDs can be reliably assigned at approximately the second birthday. The symptoms typically emerge when the individual is aged 6 to 18 months. There are no known biological markers for ASDs; the etiology of ASD is unclear.

Macrocephaly (head circumference [HC] 97th percentile) is one of the better-established phenotypic features characterizing a subset of individuals with autism. Macrocephaly (head circumference [HC] 97th percentile) is one of the better-established phenotypic features characterizing a subset of individuals with autism. Macrocephaly in autism is not apparent prenatally or at birth but emerges as the result of an abnormally rapid growth velocity in the first year of life. Neuroimaging studies suggest that enlarged HC in children with autism is, indeed, associated with increased total brain volume rather than with increased non neural tissue volume, cerebrospinal fluid, or intracranial blood volume. Enlargement in young children seems to extend to the frontal, temporal, and parietal lobes in gray and white matter and to subcortical structures, such as the amygdalae and the caudate nucleus.

Case 1

6 year old male child born by preterm delivery (36 weeks) low birth weight (birth weight of 2 kg) vaginal delivery cried immediately after birth. Child had 4 episodes of seizure (tonic clonic of upper and lower limb with clinching of teeth) from 1 year of age for which he is on oral anti epileptic drug. Head circumference at birth was 30 cm which later increased to 53 cm at 6 years of age.

O/E

Deficiency in verbal and nonverbal social communication
Limited initiation of social interaction
Difficulty coping with changes
Restricted/ repetitive behaviour appear frequently and interfere with functioning
Language milestone is delayed

Psychological assessment report— During psychological assessment good rapport was established with him. During test he was restless. Semi interview was live performance was used for assessment. His mother interview was taken.

2D echo – Normal

MRI brain plain and contrast—4.3 x 1.2 x 1.4 cm (AP X TR X CC) sized Arachnoid cyst noted in left frontal region. No obvious abnormal enhancement noted at present scan.

Weight at 6 year of life is –25 kg which is + 1 to +2 SD

Height at 6 year of life is—120 cm which is median to + 1SD

BMI at 6 year of life is -17.36 kg/m² which is + 1 to + 2 SD

Mental age – 30 months

Chronological age—72 months

Child was diagnosed as a case of strabismus with seizure with autistic spectrum disorder.

Case 2

5 year old child full term normal vaginal delivery with birth weight of 2.3 kg and head circumference of 31 cm at birth came to us with complain of seizure episodes of GTCS type lasted for 3 min which was aborted by injection midazolam

O/E

Lack of eye to eye contact
Absence of fear in presence of danger
Delayed speech
Abstract thinking is absent
Difficulty coping with changes
Restricted/ repetitive behaviour appear frequently and interfere with functioning
2D echo—normal
USG abdo pelvis – normal
NSG – normal
Weight at 5 year of life is –21 kg which is + 1 SD
Height at 5 year of life is—114 cm which is median to + 1SD
BMI at 5 year of life is -16.15 kg/m² which is + 1SD to median
Chronological age—60 months
Head circumference at 5 year of life- 51 cm
IQ range—40-54

Case 3

6 year old male child delivered by LSCS i/v/o oligohydromnios with birth weight of 2 kg and head circumference of 30 cm at birth came to us with history of seizure episode and delayed language milestone. Other milestones are with in normal range for the age group.

O/E

Lack of eye contact
Limited initiation of social interaction
Difficulty coping with changes
Restricted /repetitive behaviour
Reduced sharing of emotion/stress
Lack of facial expression
Hyperactivity
Weight at 6 year of life is –23 kg which is + 1 SD
Height at 6 year of life is—120 cm which is median to + 2 SD
BMI at 6 year of life is -15.9 kg/m² which is + 1SD to median
Chronological age—60 months
head circumference at 6 year of life- 52 cm

II. Discussion :

When considering potential underlying mechanisms for enlarged total brain volume and acceleration of head circumference growth in autism, most researchers have focused on factors that affect neuronal development. The present study, however, highlights the importance of examining factors that affect not only brain but also skeletal growth. In fact, considerable evidence suggests that many of the factors that affect neuronal growth also affect nonneural tissue and cell development. For example, fibroblast growth factor 2, which has a role in memory consolidation and neurogenesis, also affects angiogenesis, vascular remodeling, and skeletal development. Insulin-like growth factor 1 level, which has been reported to be elevated in the plasma of children with autism and is known to affect body size, is linked to brain overgrowth. Brain-derived neurotrophic factor, reported to be altered in ASDs not only has an important role in early brain development but also is associated with metabolic syndrome and obesity. Vascular endothelial growth factor, found at higher levels in plasma of adults with autism also regulates organ and body growth in early postnatal development.

III. Conclusion

Autism experienced accelerated head circumference growth in the first year of life. However, this phenomenon reflected a generalized process affecting other morphologic features, including height and weight. The study highlights the importance of studying factors that influence not only neuronal development but also skeletal growth in autism.

Dr Mohammed Arif, et. al. "Generalized Overgrowth in Autism." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 21(08), 2022, pp. 05-07.