Under-recognized surgical pathology in Autism spectrum disorder of MRI findings

Tomoko Tanaka MD; N. Scott Litofsky MD; Kristin Sohl MD; Dana Mazuru MD; Kristina J Aldridge PhD

University of Missouri School of Medicine

Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental condition characterized by impairments in social interaction, communication difficulties, and the presence of restricted, repetitive, and stereotyped patterns of behavior. According to Centers for Disease Control and Prevention the prevalence of ASD has increased from 1:150 (1992) to 1:67 (2012). Despite many research projects and advanced neuroimaging studies performed as part of ASD research, brain magnetic resonance imaging (MRI) is not recommended as part of routine evaluation, a decision supported by American Academy of Neurology, the Child Neurology Society, the American Academy of Child and Adolescent Psychiatry, and the American Academy of Pediatrics. Therefore, we wished to identify the prevalence of neuro-pathological findings in our ASD patients.

Learning Objectives

To identify under-recognized treatable cause in Autism

Methods

Retrospective electrical medical record (EMR) review of 2321 patients registered in the database of Thompson Center at University of Missouri Healthcare and Clinic between October 1990 and July 2013 was performed to identify the prevalence of MRI abnormalities in ASD patients.

Results

579 patients had brain MRI. 386 patients had normal studies. 182 patients, 31.4% (Male: female=142:40) demonstrated 208 abnormal findings. Cerebellum ecotopia(3.5%) was the most common , followed by venous anomaly (2.6%), arachnoid cyst (2.9%), Periventricular abnormality (2.1%), Chiari I malformation (1.6%),ventricle dilation/hydrocephalus(1.4%) , cortical dysplasia(0.7%), possible temporal sclerosis(0.5%), tuber sclerosis (0.18%) and three brain tumors.

Conclusions

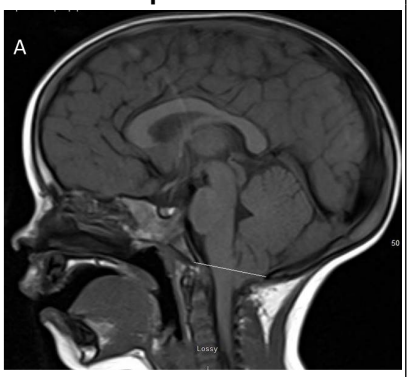
Intracranial abnormalities are relatively common in patients with ASD, suggesting that these abnormalities have likely been under-recognized. Prevalence was 10 times higher than previous report. Identifying chief complaints and performing clinical assessment of ASD patients is difficult since patients have impairment of communication. Treatable surgical pathologies like Chiari I malformation, cortical dysplasia, hydrocephalus, and arachnoid cyst, which are likely underdiagnosed in ASD patients, may affect patient functional outcome. The necessity of routine brain MRI in patients with ASD should be re-evaluated and likely be instituted.

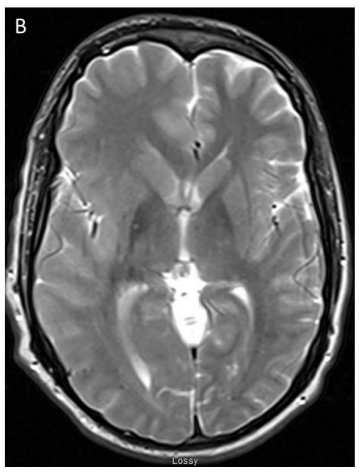
Table 1: Prevalence of result compared to general population

MRI finding	Prevalence of result (%)	General Prevalence (%)
Cerebellum Ectopia	3.35	*
Venous angioma/anomaly	2.64	0.96
Arachnoid cyst	2.29	2.60
Periventricular abnormality	2.11	*
Chiari I Malformation	1.58	0.77
Leukomalacia	1.41	*
Ventricle Dilation	1.41	*
Agenesis Corpus Callosum	1.41	0.03-0.05
Pineal cyst	1.06	1.98
Cortical dysplasia	0.70	*

*: There was no published general prevalence

Brain MRI Eample of Autism patients





A: T1 Midsaggital demonstrates cerebellar tonsile distension 10mm (white line)Chiari I malformation,B: T2 Axial view right entire hemispheric enlarged consists with hemimegancephaly.

References

- 1.Zeglam A. MRI or not to MRI! Should brain MRI be a routine investigation in children with autistic spectrum disorders? Acta Neurol Belg. 2015;115:351-354.
- 2.Jayarao M, Sohl K, Tanaka T. Chiari malformation I and autism spectrum disorder: an underrecognized coexistence. J Neurosurg Pediatr. Jan 2015;15(1):96-100.
- 3.Filipek PA, Accardo PJ, Ashwal S, et al. Practice parameter: screening and diagnosis of autism: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Child Neurology Society. Neurology. Aug 22 2000;55(4):468-479.
- 4. Johnson CP, Myers SM, American Academy of Pediatrics Council on Children With Disabilities: Identification and evaluation of children with autism spectrum disorders. Pediatrics 120:1183–1215, 2007
- 5. Volkmar F, Cook EH Jr, Pomeroy J, Realmuto G, Tanguay P: Practice parameters for the assessment and treatment of children, adolescents, and adults with autism and other pervasive developmental disorders. J Am Acad Child Adolesc Psychiatry 38 (12 Suppl):32S–54S, 1999