

PREVALENCE OF MINOR CONGENITAL ANOMALIES IN CHILDREN WITH HYPERACTIVITY

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Abstract. A congenital anomaly is a physical, metabolic, or anatomic deviation from the normal pattern of development that is present at birth. Minor congenital anomalies do not have medical or cosmetic importance, but detection of more than three such anomalies may reveal the prenatal origin of a disorder. The aim of the present study was to establish an association between the presence of minor congenital anomalies and hyperactivity, learning disabilities and attention deficit. 219 patients aged between 7 and 18 years were examined for the presence of minor congenital anomalies and compared to a control group. The frequencies of these anomalies were not significantly different in the two groups. The mean values of minor congenital anomalies per child were significantly different: 2.5 in children with learning disabilities, 4 in those with learning disabilities and attention deficit and hyperactivity, 1.05 in the control group. Prevention is the best approach to congenital anomalies. As a positive correlation could be established, it was concluded that the presence of minor congenital anomalies especially more than three, may predict the future onset of these disorders.

Keywords: minor congenital anomalies, hyperactivity, predictive value

INTRODUCTION

A congenital anomaly is a physical, metabolic, or anatomic deviation from the normal pattern of development that is present at birth. Minor congenital anomalies do not have medical or cosmetic importance, but detection of more than three such anomalies may reveal the prenatal origin of a disorder. They may occur even in healthy individuals. The incidence of minor anomalies has been studied in search for a relationship between different non-syndromic diseases and abnormal embryogenesis. They are used as indicators of altered embryonic differentiation. Their positive correlation with disturbances of neurological development occurring during embryogenesis has been established. Patients with mental retardation, cerebral palsy, autism and other neurological disorders were evaluated for the presence of minor anomalies [1-4]. The presence of minor congenital anomalies, especially more than three, may predict the future onset of a neurological disorder.

Attention Deficit Hyperactivity Disorder (ADHD) is one of the most common mental disorders that develop in children, who have impaired functioning in multiple settings, including home, school, and in relationships with peers. It is a neurobehavioral developmental disorder estimated to affect about 3-5% of the world's population under the age of 19 [5]. It typically is diagnosed during childhood, and is characterized by a persistent pattern of inattention and/or hyperactivity, as well as forgetfulness, impulsivity, and distractibility. It appears to be highly heritable, although one-fifth of all cases are estimated to be caused from trauma or toxic exposure. Evidence suggests that hyperactivity has a strong heritable component, and in all probability ADHD is a heterogeneous disorder, meaning that several causes could create very similar symptomology [6].

Learning disabilities are problems that affect the brain's ability to receive, process, analyze, or store information [7]. The causes for learning disabilities are

not well understood, and sometimes there is no apparent cause for a learning disability [8]. However, some causes of neurological impairments include:

- Heredity - Learning disabilities often run in the family
- Problems during pregnancy and birth
- Accidents after birth

The aim of the present study was to establish an association between the presence of minor congenital anomalies and hyperactivity, learning disabilities and attention deficit.

MATERIALS AND METHODS

219 subjects aged between 7 and 18 years were examined for the presence of minor congenital anomalies and compared to a control group. The incidence and the mean number of minor congenital anomalies were investigated in patients hospitalized in the Neuropsychiatry Center for children and Teenagers from Timisoara. Data were collected by performing multiple physical examinations and measurements where necessary for a greater accuracy [9]. Minor anomalies studied are presented in table 1.

A full history was taken, with questions about prenatal, perinatal and postnatal period. Data regarding the age of the parents, mother's age being of special interest, regarding pregnancy including illnesses, drugs or use of alcohol during this time were collected. Other important data were those concerning length of pregnancy, child's weight at birth, the existence of twin pregnancies, as well as the existence of spontaneous abortion or stillbirth in the family. Perinatal history included data about labour, birth, presentation, Apgar score and the condition of the child during the first week of life. Data about postnatal history regarded the psycho-motor development of the child, existence of trauma, severe disorders during infancy or childhood or surgery [6, 7].

Table 1. Minor congenital anomalies

Face	Synophris, anteverted nostrils, bifid tip of nose, high arched palate, bifid uvula, micrognathia,
Eyes	Epicanthic folds, upslanting palpebral fissures, downslanting palpebral fissures, short palpebral fissures, hypertelorism, hypotelorism
Ears	Malformed ears, asymmetric ears, low set ears, small ears, preauricular pits or tags, ear lobe creases
Head and Neck	Webbed neck, flat occiput
Hair	Two or more parietal whorls, abnormal posterior whorl
Hands	Clinodactily, partial cutaneous syndactily, simian crease, Sydney crease,
Trunk	Accessory nipples, short stern, haemangioma, cafe-au-lait spots
Feet	Broad hallux, partial syndactily

The control subjects consisted of children and teenagers corresponding as ages and sex, with no history of neurodevelopmental disorders. They were chosen randomly from kindergartens and schools.

RESULTS AND DISCUSSIONS

Establishing of a positive association with some markers could be useful for identification of the cause and the prenatal or postnatal onset of these disorders. These markers could be represented by minor congenital anomalies.

Sex ratio in affected children was:

- Boys: 65%
- Girls: 35%.

The age of the patients varied between 3 and 18 years, the mean age being 11.35 years.

The incidence of minor congenital anomalies in the autistic group was 14.47%, approximately equal to their incidence in the control group, 14.8%. The frequencies of these anomalies were not significantly different in the two groups. The mean values of minor congenital anomalies per child were significantly different: 2.5 in children with learning disabilities, 4 in those with learning disabilities and attention deficit and hyperactivity, 1.05 in the control group.

About 85% of the minor congenital anomalies seen in these patients are anomalies of the mouth, eyes, and ears. The minor anomalies, most frequently found were:

- high arched palate
- epicanthus
- hypertelorism
- malformed ears
- upslanting palpebral fissures

Regarding the anomalies of the hand, only clinodactily of the fifth digit was seen.

Anomalies of the limbs were found in a proportion of 14.70%.

The anomalies frequently seen were:

- clinodactily of the fifth digit, unilateral or bilateral,
- anomalies of the toes,
- modified dermatoglyphics.

Studying the histories of the patients, it was noticed that:

- almost half of the affected children had very young mothers or the age of the mothers at conception was over 34 years. Thus, 23.8% of the children had very

young mothers (aged between 17 and 20) and other 23.8% had mothers that were between 34 and 41 at conception.

- 30% of the children mentioned above also had major congenital defects
- one of the pregnant women used alcohol during pregnancy, the child presenting signs of the fetal alcohol syndrome.

Postnatal history did not reveal significant data for this study.

Minor congenital anomalies can be an important tip-off that a neurologic disorder in childhood is caused by a congenital syndrome rather than a problem that occurred during labor and delivery. Considering the clinical manifestations of the disorders, that require a complex sustained and long duration treatment, establishing of an early diagnosis is absolutely necessary [10]. It's therefore critical that a child be examined by a clinical geneticist or dysmorphologist. It is estimated that about 10% of children with two minor malformations have a major abnormality, such as a cardiac malformation or a neurologic disorder, such as mental retardation, developmental delay seizures, spasticity, and psychosis. About 20% of children with three or more minor malformations have a major abnormality. The observations from this study, more than half of the patients having three or more minor congenital defects lead to the conclusion that a careful examination of the infant or child, for establishing the presence of multiple minor congenital defects in one patient, might represent a marker of a central nervous system developmental dysfunction. Detailed and accurate clinical examination may help establishing genotype-phenotype correlations.

Prevention is the best approach to congenital anomalies. As a positive correlation could be established, it was concluded that the presence of minor congenital anomalies especially more than three, may predict the future onset of these disorders.

REFERENCES

- [1] Tenyi T, Trixler M, Csabi G, Jeges S. Minor physical anomalies in non-familial unipolar recurrent major depression. *J Affect Disord.* 2004 Apr; 79 (1-3):259-62.
- [2] Mehes K, Kosztolanyi G. Genetic evaluation of mental retardation. *Orv Hetil.* 1998 Feb 15;139 (7):339-46.
- [3] Megan L Wier, Cathleen K Yoshida, Roxana Odouli, Judith K Grether, Lisa A Croen. Congenital anomalies

- associated with autism spectrum disorders. *Developmental Medicine & Child Neurology* (2006), 48: 500-07
- [4] Rebecca Muhle, Stephanie V. Trentacoste, Isabelle Rapin. The Genetics of Autism. *Pediatrics* Vol. 113 No. 5 May 2004, 472-86
- [5] Polanczyk G, de Lima MS, Horta BL, Biederman J, Rohde LA (2007). "The worldwide prevalence of ADHD: a systematic review and meta-regression analysis". *Am J Psychiatry* 164 (6): 942–48.
- [6] Roman T, Rohde LA, Hutz MH. (2004). "Polymorphisms of the dopamine transporter gene: influence on response to methylphenidate in attention deficit-hyperactivity disorder." *American Journal of Pharmacogenomics* 4(2):83–92
- [7] Margai, F. & Henry, N. (2003). A community-based assessment of learning disabilities using environmental and contextual risk factors. *Social Science & Medicine*, 56(5), 13.
- [8] Patti L. Harrison; Flanagan, Dawn P. (2005). *Contemporary intellectual assessment: theories, tests, and issues*. New York: Guilford Press.
- [9] Judith H. Miles, Richard E. Hillman. Value of a Clinical Morphological Examination in Autism. *Am J Med Genetics* 91, 2000, 245-53
- [10] Harper PS. *Practical Genetic Counselling*, 5th Edition. Boston: Butterworth Heinemann, 1998: 11, 56-70.