

Photoanthropometric Study of Dysmorphic Features of the Face in Children with Autism and Asperger Syndrome

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Objective: Childhood autism is a neurodevelopmental disorder characterized by impairments in social interactions, verbal and non-verbal communication and by a pattern of stereotypical behaviors and interests. The aim of this study was to estimate the dysmorphic facial features of children with autism and children with Asperger syndrome .

Methods: The examination was conducted on 60 children (30 with childhood autism and 30 with Asperger syndrome). The photo anthropometric method used in this study followed the protocol established by Stengel-Rutkowski et al .

Results: The performed statistical analysis showed that in patients with childhood autism, the anteriorly rotated ears and the long back of the nose appeared more often. In the group of children with autism, there was a connection between the amount of dysmorphies and the presence of some somatic diseases in the first-degree relatives. There was also a connection between the motor coordination and the age the child began to walk.

Discussion: In patients with childhood autism, there were certain dysmorphies (like the anterior rotated ears and the long back of the nose) which appeared more often. Although the connection was not statistically significant, it seemed to concur with data from the literature .

Conclusion: Formulation of the other conclusions would require broader studies e.g. dealing with a familial analysis of dysmorphic features.

Keywords: *Autistic Disorder, Asperger syndrome, Etiology, Feature*

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Childhood autism is a disorder which, in spite of its discovery over 60 years ago, still remains a scientific challenge. Although studies show significant genetic and neurobiological factors in its pathogenesis, the aetiology of autism has not yet been established. Its biological causes have not been discovered, nor has a defined constant marker for it ever been found (1). At present the prevalence of autism is estimated at 1-2 per 1000 children, and autism spectrum disorders (ASD) at 3-6 per 1000 (1, 2, 3, 4, 5, 6). Autism is not a pathogenically homogenous syndrome; it is characterized by disorders of structure and activities of the central nervous system, which can be the cause of the impairments in social interactions, communication and a pattern of stereotypical behaviours and interests.

In contrast to the majority of children with disorders that are associated with mental retardation, autistic children are generally regarded as very attractive. Statements regarding this can be found in work by Hans Asperger, who emphasized the symmetrical features of autistic persons' faces, describing them as "aristocratic, even slightly degenerate" (7). The problem of the head and body build of persons with

autism has very rarely been taken up in other studies. Certainly the fact that such persons do not tolerate physical contact, reacting with loud shouts and protests, has had a major influence on this, and that is why it has been difficult to perform anthropometric measurements which require very close contact. Therefore, we decided to use indices calculated on the basis of comparing the parameters on facial photographs instead of performing direct anthropometric measurements .

The aim of this study was to estimate the dysmorphic features of the face of children with childhood autism and children with Asperger Syndrome (AS).

Materials and Method

This The study was performed in a group of 60 children (30 with childhood autism and 30 with Asperger Syndrome). Criteria DSM-IV (8) was used in medical examination and diagnosis, and the Gillberg criteria for Asperger Syndrome was also used (9). The medical exam and diagnoses were undertaken in a psychiatric out-patient clinic by two psychiatrists and two pediatricians. The somatic and psychiatric status of the children's family was determined through interview with parents .

All children were psychiatrically examined and their behavior in new surroundings was observed. The criterion for exclusion in the study was the presence of any chromosomal defects manifesting phenotypically, such as Down syndrome. The photo anthropometric method used in this study followed the protocol established by Stengel-Rutkowski et al. (10). This method was successfully used in different studies (11, 12, 13). The obtained data were compared to the norms of this scale (10, 14). (Table 1).

The values below the 3rd and above the 97th percentile were defined as dysmorphic. Additionally, parents were asked to complete a short questionnaire check-list to broaden the anamnestic data. Fifty one questionnaires were received; some parents did not answer all the questions.

Statistical analysis was conducted using ANOVA Kruskal-Wallis and U Mann Whitney tests. Correlation analysis was done using the χ^2 test and the Spearman index. A level of $p < 0,05$ was accepted as statistically significant. Calculations were conducted using "Statistica 5.0 Pl" software (StatSoft INC., USA).

Results

We examined 52 boys and 8 girls, aged between 3 and 12 years. In the group of children diagnosed with childhood autism, there were 25 boys and 5 girls (the proportion of boys to girls was 5:1) ages 3-12 (mean: 7.1; SD=2.604). In the group of children diagnosed with Asperger Syndrome, there were 27 boys and 3 girls (the proportion of boys to girls was 9:1) ages 5 to 12 (mean: 9.483; SD=2.325). Both groups were homogeneous as for sex; however, they differed as for age. The number of dysmorphies in the group diagnosed with autism was from 0 to 6 (mean: 2.1), whereas the number was from 0 to 4 (mean: 1.933) in the group with Asperger Syndrome. Both groups were homogeneous in the number of dysmorphies ($p=0.6034$). Most of the children diagnosed with autism were characterized by having good motor coordination, whereas most of the children with Asperger Syndrome were characterized by having poor

motor coordination. Motor development (the onset of walking) and development of speech (first words) of children in both groups are presented in Table 2.

The examined groups did not differ in a statistically significant way concerning the time of the appearance of abnormal behavior, so their parents did not notice anything unusual in the children's behavior. In all children diagnosed with childhood autism such behaviour was observed before they reached 4 years old, whereas in 25% of the children with Asperger Syndrome, this was noticed after they reached the age of 4 (Table 3).

Serious somatic illnesses occurred in the first-degree relatives of 12 (52.17%) of the persons in the study group (cancer, diabetes, asthma, epilepsy, stomach ulcers, ulcerative colitis, Duchenne muscular dystrophy, Parkinson's disease, multiple sclerosis) as well as in 14 (66.67%) of the persons in the Asperger group (malignant tumors, Parkinson's disease, hypothyroidism, systemic lupus erythematosus, stomach ulcers, asthma, diabetes). Mental illnesses of first-degree relatives occurred in 6 (27.27%) of the children diagnosed with autism (schizophrenia, depression, alcohol dependence syndrome) and 10 (43.48%) of the children with Asperger Syndrome (schizophrenia, depression, obsessive-compulsive disorder, alcohol dependence syndrome, mental retardation). The performed statistical analysis showed that in patients with childhood autism the anteriorly rotated ears and the long back of the nose appeared more often (Table 4)

In the group of children with Asperger Syndrome, there was a relationship between the amount of dysmorphies and sex ($p=0,0350$); females were found to have a larger number of dysmorphies. In the group diagnosed with autism, a connection was found between the amount of dysmorphies and occurrence of serious somatic illnesses in the first-degree relatives. The occurrence of these diseases was connected with a greater number of dysmorphies. In the group diagnosed with childhood autism, there were also connections (approaching statistical significance) between the

Table 1. The values of photoanthropometric indices in the examined groups*.

Index	Mean	Min.	Max.	SD	The coefficient of variation [%]
1. Midface height	0,597	0,48	0,69	0,05	8,37
2. Inner canthal distance	0,241	0,16	0,30	0,02	8,30
3. Width of the palpebral fissures	0,204	0,17	0,24	0,015	7,35
4. Length of the back of the nose	0,462	0,33	0,58	0,05	10,82
5. Interalar distance	0,250	0,19	0,30	0,215	8,60
6. Prominence of the upper jaw	0,942	0,83	1,05	0,04	4,25
7. Nasolabial distance	0,171	0,11	0,22	0,03	17,54
8. Width of the mouth	0,333	0,28	0,42	0,03	9,00
9. Height of the chin	0,211	0,14	0,30	0,03	14,22
10. Prominence of the chin	1,027	0,92	1,26	0,07	6,81
11. Inclination of the ear insertion line	79,983	64,00	97,00	6,61	8,25
12. Length of the ears	0,754	0,59	0,86	0,05	6,63
13. Width of the ears	0,334	0,20	0,41	0,04	11,98

* The values of photoanthropometric indices are established by Stengel – Rutkowski et al 1984)

Table 2. Motor and speech development in the examined groups

First steps [age in months]	Childhood autism [n]	Asperger Syndrome [n]
8-12	12	7
13-15	8	11
16-18	3	5
19-24	1	2
>24	2	0
First words [age in months]	Childhood autism [n]	Asperger Syndrome [n]
8-12	12	15
13-15	-	4
16-24	5	2
24-36	4	1
36-48	2	2
>48	1	1
Lack of speech	2	0

Table 3. Appearance of abnormal behaviour in the examined groups

DIAGNOSIS	Time of the abnormal behaviour appearance [age in months]							Together
	0-3	4-6	7-12	13-24	24-36	36-48	>48	
Childhood autism	3	4	3	5	8	3	0	26
Asperger Syndrome	3	3	3	4	0	5	6	24
All	6	7	6	9	8	8	6	50*
X ² Pearson	14,69748		Df=6		p=0,02275			

*Not all parents gave answers in inventory

Table 4. The difference in the frequency of dysmorphism appearance in the examined groups.

Index	% of dysmorphism for the group with childhood autism autizm		% of dysmorphism for the group with AS		Difference in the frequency of dysmorphism appearance [n] level	
	>97 th centile	<3 rd centile	>97 th centile	<3 rd centile	>97 th centile	<3 rd centile
1. Midface height	13,33	6,67	0	16,67	p=0,11	p=0,42
2. Inner canthal distance	3,33	3,33	0	0	p=1	p=1
3. Width of the palpebral fissures	26,67	0		3,33	p=0,14	p=1
4. Interalar distance	6,67	10,00	10,00	10,00	p=1	
5. Width of the mouth	3,33	0	16,67	0	p=0,19	
6. Length of the back of the nose	23,33	3,33	0	0	p=0,01	p=1
7. Prominence of the upper jaw	0	3,33	0	13,33		p=0,35
8. Nasolabial distance	0	6,67	3,33	0	p=1	p=0,49
9. Height of the chin	0	30,00	3,33	30,00	p=1	
10. Prominence of the chin	3,33	6,67	16,67	10,00	p=0,19	p=1
11. Inclination of the ear insertion line	0	40,00	0	13,33		p=0,04
12. Length of the ears	3,33	6,67	6,67	0	p=1	p=0,49
13. Width of the ears	6,67	0	6,67	6,67	p=1	p=0,49

number of dysmorphies and motor coordination (p=0,076; a greater number of dysmorphies had a connection with poor motor coordination) and between the number of dysmorphies and the beginning of walking (p=0,1318).

In the group of children diagnosed with Asperger Syndrome, there was a relationship between the amount of dysmorphies and the age the child articulated its first words (p=0,0309).

Discussion

In this study, among children diagnosed with autism, the number of boys was five times more than girls. This proportion is in accordance with epidemiology data, which can confirm the representativeness of the study group (15). This large predominance of males is

currently believed to be the result of a higher level of fetal testosterone. Its role in the pathogenesis of autism may be found, among other things, in the so-called exposure index for fetal testosterone (2D:4D ratio – the second to the fourth digit), which is lower in persons with autism, and also in a higher level of androgens and the early maturity of adolescents with autism compared with those in the control group (16, 17). The role of testosterone in the pathogenesis of autism is emphasized in the extreme male brain theory by Simon Baron-Cohen (18).

In the group of children diagnosed with Asperger Syndrome, the number of boys was ten times more than girls. Such a large predominance of boys is most likely connected with age, when it is more difficult to diagnose Asperger Syndrome in girls who, as a rule, are characterized by a higher level of social abilities.

As demonstrated in the attached bibliography, among children with AS sent for diagnosis, the proportion of boys to girls is 10:1 (19).

Furthermore, in the group of children diagnosed with AS, a connection between the amount of dysmorphies and sex was observed. There were more dysmorphies among females, which could confirm the existence of deeper disorders in girls. This concurs with data of other studies, according to which disorders of neurobiological nature occur more frequently in boys, whereas in girls they result in deeper developmental problems. The threshold of susceptibility for the damaging factors is lower in boys, whereas the occurrence of disorders is preceded by a greater amount of destructive factors in girls. Hence, the disorders in girls are more intensive and more frequently connected with cognitive impairment (20).

The majority of children with autism, in regard to physical development, are characterized by proportionate body build and proper height (21, 22). Nevertheless, different authors describe certain dysmorphic features, commonly occurring singly in autistic persons (23).

In the examined groups of autistic children, there were certain small anomalies like the anteriorly rotated ears ($p=0,04$) and the long back of the nose ($p=0,01$) which appeared more often, but the connection was not statistically significant. In other studies, the occurrence of minor physical anomalies (MPAs) in neurological disorders such as schizophrenia, ADHD, fetal alcohol syndrome and cerebral palsy are often described (24, 25). Since the minor physical anomalies are connected more often with structures deriving from the ectoderm, from which the nervous system develops, their presence might indicate its incorrect development, appearing most likely during the first or at the beginning of the second trimester of pregnancy. One can suppose that anatomical anomalies interact with other genetic and environmental factors in creating the symptoms of the illness. The present state of medical knowledge allows us to establish the precise time of the genesis of the concrete deformations, as concrete organs develop in a definite period of pregnancy. Stromland et al. (2002) in their work dealing with children diagnosed with autism, whose mothers took thalidomide during pregnancy, it was established that the majority of these children had anomalies in their external ears as well as impairments in their facial nerves VI and VII (Moebius syndrome) and proper developed limbs (26). Most likely autism with Moebius syndrome is connected with using thalidomide between the 20th and the 24th days of pregnancy (26). Minor physical anomalies are described as well in other cases of childhood autism, among them, hypotelorism or numerous abnormalities concerning ears – such as, for example, posterior rotation of the external ears, underdevelopment or lack of earlobe, the low position of auricles (22, 27). Casas et al (2004) described dysmorphies of the ear as well as nose in their work concerning the autistic patients with deletion 2q (28).

In the group diagnosed with childhood autism, there was a connection between the number of dysmorphies and motor coordination (a greater number of dysmorphies had a connection with poor motor coordination) as well as when autistic children begin to walk. The motor development of children with autism rarely has been the subject of studies; a motor pattern characteristic for autism has not yet been established. Johnson et al (1992) stated that motor development was distinctly delayed in about 28% of children. In a study by Teitelbaum et al, (1998) all infants whose autism was later diagnosed were found to have certain abnormalities in motor development (29, 30). Delayed motor development may be connected with perinatal complications, additional somatic illnesses or mental retardation. It is likely that such disturbance in child development worsens its adaptability. A greater amount of dysmorphies seems to be connected with a greater degree of developmental disorder.

In our study, we also found that the both examined groups of children differed in the time of the appearance of their abnormal behavior in a statistically significant way, so their parents did not notice anything unusual about their behavior. In 25% of the children with Asperger Syndrome, such behaviour was observed after they reached the age of 4. These results are in accordance with that of other studies. Certain symptoms of Asperger disorder may be present very early, however, because children with AS have better social contact than autistic children, diagnosis is established later - specific difficulties in social interactions as well as narrow and limited interests became more noticeable when the child began school education (31, 32, 33). The latest studies show that the average age of a child with Asperger Syndrome at the time of diagnosis is 11 years old (whereas for a child with autism it is 5.5 years old) (34, 35).

In the studies conducted on children diagnosed with childhood autism, there was a connection between the amount of dysmorphies and the appearance of serious somatic illnesses in the first-degree relatives. The presence of illness in the family can be an indicator of a greater risk for the next child or its greater susceptibility to damaging factors. Data from other studies show a statistically more frequent occurrence of certain diseases among the relatives of persons with autism – breast and uterus cancer among mothers as well as auto-immunological illnesses (diabetes type I, ulcerative colitis, hypothyroidism, rheumatoid arthritis, psoriasis as well as systemic lupus erythematosus) among mothers and first-degree relatives (36, 37, 38, 39). In other studies, one can find data relating to the existence of certain immunological abnormalities in autistic persons (40). However, formulation of eventual conclusions about the connection between the presence of auto-immunological illnesses among relatives and the occurrence of childhood autism requires further studies.

In the presented material in the group with Asperger Syndrome, there was a relationship between the

amount of dysmorphies and the development of speech. A greater number of dysmorphies was connected with the later age at which a child articulated its first words. This connection requires further studies. Our study has some limitations. To eliminate the anomalies running in a family it would be useful to also examine the parents and siblings of the autistic children. We also considered extending our study for the control group but we finally decided to use the standards established by Stengel-Rutkowski et al. from white control children.

We conclude that in the group of children with autism, there was a connection between the amount of dysmorphies and the presence of some somatic disorders in the first-degree relatives. The number of dysmorphies also showed the connection with motor coordination and the age the child began to walk.

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