

**SYMPOSIUM**

## Body shape of patients with chromosomal aberration<sup>†</sup>

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**ABSTRACT** The body shape of ten patients (4 with different forms of mucopolisaccharidosis, 2 with Cockayne syndrome and 1-1 with Klippel-Feil, Sturge-Weber, Rubinstein-Taybi and Cri du chat syndrome) are described in this study. Our aim is to call the attention to the importance of the detailed antropological examination of these patients, because this is one of the possible ways to understand how different genes influence of the shape of the body.  
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**KEY WORDS**

chromosomal aberration  
syndromes  
body shape  
proportionality

One of the ways to understand the genetic effects on the body shape and the growth is to study the body dimensions in monogene disorders and chromosomal disorders. The number of such publications is limited because of two difficulties. First is the rarity of these syndromes: it is practically impossible to gather enough data for statistical analysis. The other difficulty in this work is that there is no generally used anthropometrical protocol available. The description of the syndromes in the literature used contains some body measurements of the given subject, but these are not enough for a detailed analysis of the body shape. Mainly general remarks on the body shape are given as “backwardness of growth” or “craniofacial dysmorphism”.

The influence of the X and Y chromosomes (Ross et al. 1983; Varrela 1984; Varella and Alvesalo 1984; Eiben et al. 1985) and also the chromosome 21 – at least the distal part – on the growth and body shape (Buday 1990) are better known because the different anomalies of the sex chromosomes, and also the trisomy and the translocation forms of the Down’s syndrome, are more frequent.

Some of these syndromes involve normal intelligence but mental retardation is found among the features most of the time. There are more than 500 genes which are able to cause mental retardation. Their effects on the body shape are not known.

### Materials and Methods

We present the body shape of ten patients living in different institutions and nursing homes in Hungary. Some of these syndromes are metabolic disorders, caused by one gene. It means that the product of this gene is missing from a metabolic chain. Therefore, the given chain changes its direction

and the by-products have an influence on the brain and/or the mental capacity, and also on some other obligatory or facultative features. So it has pleiotropic effects including an influence on the growth and the body shape because of the disturbed metabolic chain. We have some other patients with structural disorders of different chromosomes.

In the following descriptions, some characteristic features are summarized, mainly which concern the growth and body shape. Then, the proportionality profile is shown. The somatotype components were also calculated for all subjects with the aim of studying the physique. The distance between the individual somatoplots and the control ones of the same age and sex, the somatotype attitudinal distance were calculated according to Duquet and Hebbelinck (1977). The control data were calculated from the Nationwide Growth Study (Eiben et al. 1991).

### Results and Discussion

#### Mucopolysaccharidoses

This is a group of diseases called lysosomal storage disorders. It has ten different forms and subforms due to the lack of different enzymes in the metabolism of glycosaminoglycans. We have four cases in this syndrome.

#### *Patients 1 and 2 on MPS IV. (Maroteaux-Lamy syndrome)*

Backwardness of the growth, craniofacial dysmorphism, convex sternum, lumbal kyphosis and scoliosis. Hard of hearing. Recessive inheritance. Locus D:12q14, McKusick number is 252900. The most frequent form of MPS, frequency is: 1:25,000.

Our patients are a 10-year-old dizygotic twin pair. Almost all of our boy patients’ body measurements are proportionally lower than that of the control (Fig. 1). There are some exceptions: length of the extremities and the humerus and femur width. The proportion of the girl pair is similar (Fig.

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<sup>†</sup>Dedicated to Professor Gyula Farkas on the occasion of his 70<sup>th</sup> birthday.

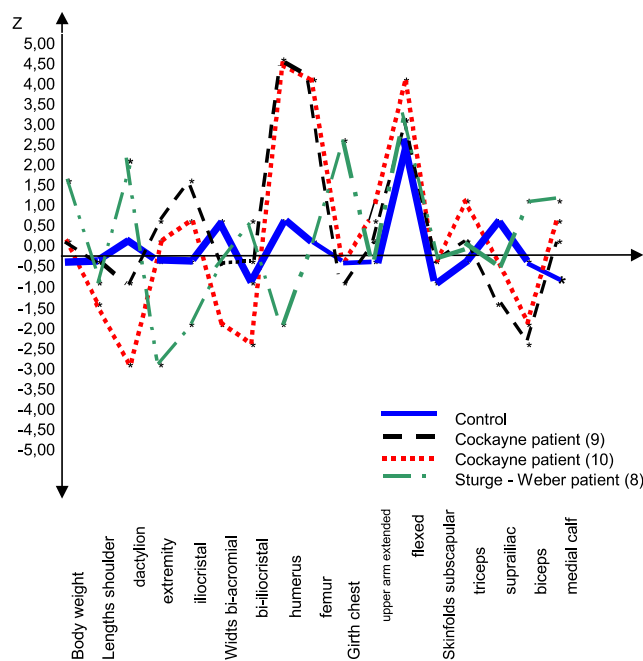


Figure 1. Proportionality profile of 10-year-old boy patients.

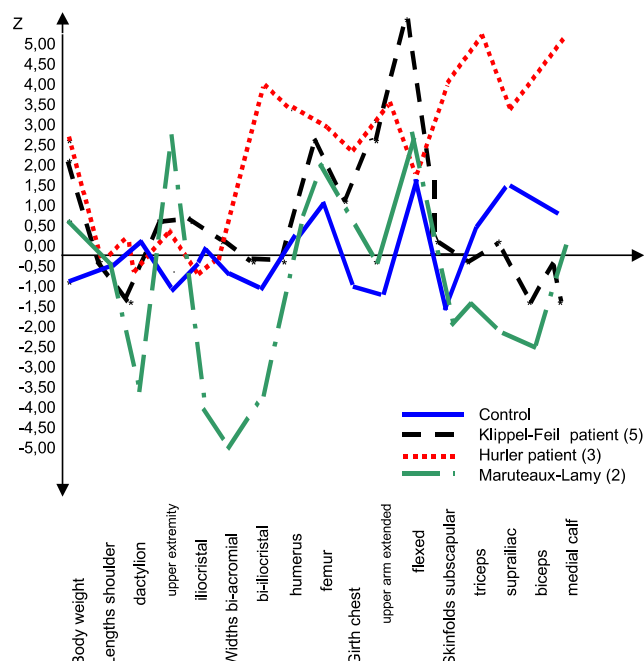


Figure 2. Proportionality profile of 10-year-old girl patients.

2) but the length of the lower extremity is proportionally lower than that of the control.

*Patient 3. MPS I. (Hurler syndrome)*

In this form, the  $\alpha$ -I-iduronidase enzyme is absent. Mental retardation and hearing impairment due to the quantity of ganglioside increase in the nervous system. Imbecillity. The “gargoil” face, short stature and deformity of skeleton (thoracolumbal gibbus, pectus carinatum, macrocephalia) are characteristic. Locus: 4p16.3, McKusick number is 252800, incidence: 1:25,000.

We have a 10-year-old girl, her proportionality profile is shown in Figure 2. The weight, girths, skinfolds and the bipectondylar width are proportionally higher than that of the controls of the same gender and age.

*Patient 4. MPS III. (Sanfilippo syndrome)*

Sanfilippo is the most frequent form of the MPS, frequency is 1:25,000. There are four different subforms in this syndrome. Growth is retarded, the features often characterized only after the second year of life. Head girth is higher than the normal probably due to the thick cranial bones. Recessive inheritance.

Our case is a Sanfilippo A form, which means the lack of heparan-N-sulfatase enzyme. McKusick number is 252900.

He is 10 years old and his lengths and widths and all skinfolds are proportionally lower than that of the control of the same age (Fig. 1).

*Patient 5. (Klippel-Feil syndrome)*

Main features are craniofacial dysmorphism, short neck with limitation of motion due to the fusion of cervical vertebrae and atlas assimilation. Frequent association with other syndromes. Autosomal dominant inheritance.

Incidence is 1:50,000, more in females. Locus is not known. McKusick number is 148900.

Our patient is a 10-year-old girl (Fig. 2). Her lengths and the biacromial and iliocrystal widths are proportionally less than that of the controls of the same gender and age. The humerus and femur widths and the girths are somewhat higher than that of the control but the skinfolds are lower. There are significant differences in the body proportion of this girl and the two other cases with mucopolysaccharidosis.

*Patient 6. (Cri du chat syndrome)*

The main feature is that the sound of the crying of these children is similar to the cat mew, because of the developmental disorder of the laryngs. Microcephaly, hypertelorism, lip and/or cleft palate.

This is a deletion: the 5p15 monosomy. Most of the cases are de novo deletion. Prevalency at birth is 1/50,000 but the frequency among mentally retarded is 1.5/5,000. Male-female ratio is 1:4.

Our patient is a 7-year-old boy. Weight, length of his extremities, the condylus width and the girths are proportionally higher than that of the control of the same sex and age (Fig. 1). There are also significant differences between the

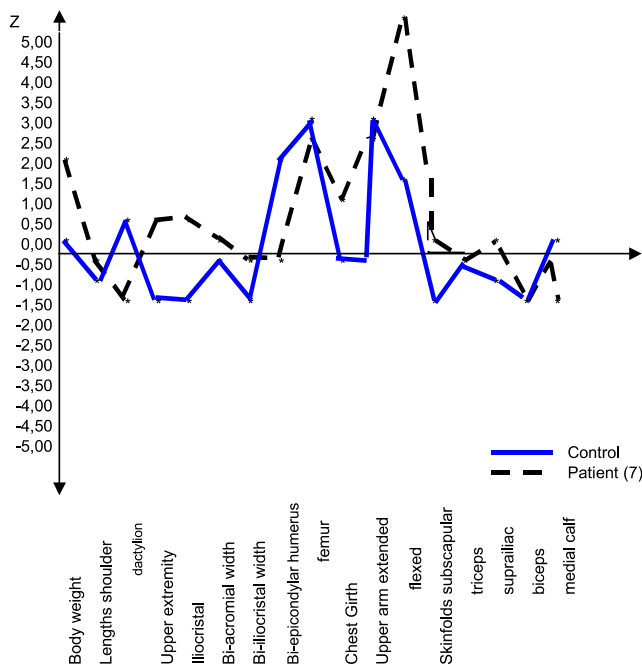


Figure 3. Proportionality profile of 5-year-old female patient with Rubinstein-Taybi syndrome.

proportion of this boy and the two other same-age patients with mucopolysaccharidosis.

Patient 7. (Rubistein-Taybi syndrome)

Main features of this syndrome are the short stature, craniofacial dysmorphism with beak-like nose, hypertelorism, microcephaly, epicanthus, maxillar hypoplasia, gothic palate, characteristic deformation of the fingers and toes (brachymegalophalangia). Imbecillity. Facultative micrognathia, pectus excavatum or carinatum, skeletal age is delayed. Autosomal dominant inheritance, locus: 16p13.3, McKusick number: 180849. Not rare.

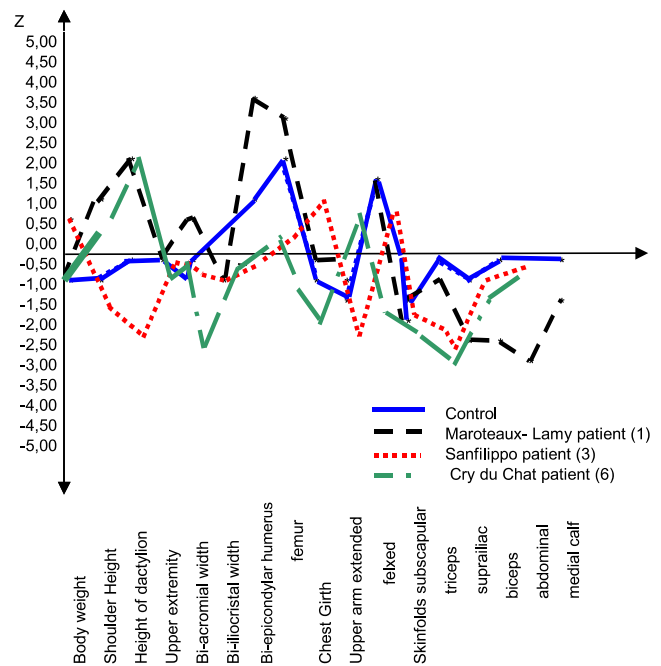


Figure 4. Proportionality profile of patients No 8, 9 and 10.

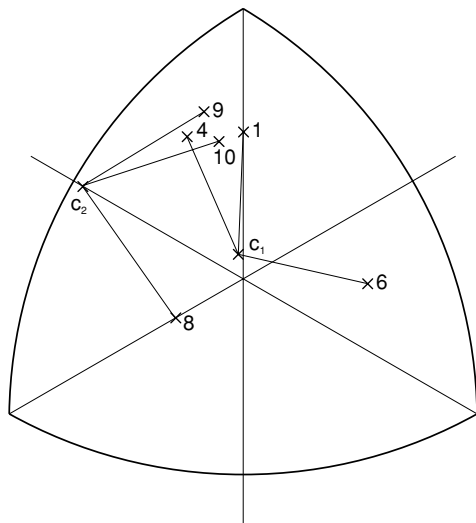
Our patient is a 5-year-old girl (Fig. 3). Her extremities and also the biacromial and bi-iliocristal width are proportionally longer than that of the control.

Patient 8. (Sturge-Weber syndrome)

This syndrome is characterized by the cutaneous angiomatosis which is unilateral on the area of nervus trigeminus and sometimes of the neck, trunk and extremities. Imbecility and focal epilepsy due to the haemangiomas extended to the leptomenings. Contralateral hemiparesis is frequent. Retardation of the growth. Mendelian inheritance is not proven although some cases with dominant inheritance were de-

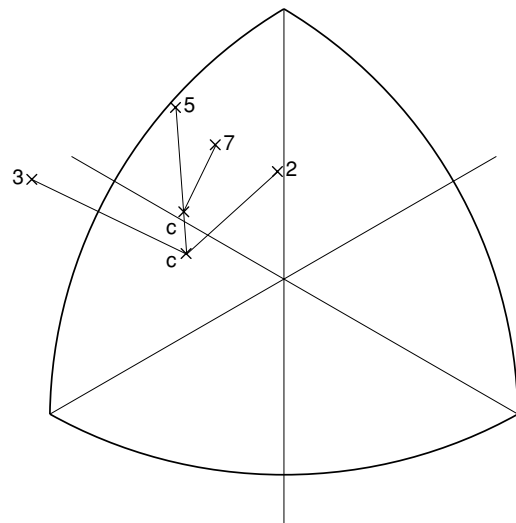
Table 1. Somatotype components of the patients.

Number	Age	Sex	I	II	III	SAD
7.	5	Female	2.00-	4.00-	0.50	2.79
Control			3.99-	4.98-	2.19	
1.	10	Male	1.00-	5.00-	1.00	3.83
4.	10	Male	3.00-	5.50-	0.50	2.79
6.	10	Male	1.00-	2.50-	4.00	3.15
Control			3.67-	4.17-	3.61	
2.	10	Female	1.50-	4.00-	1.50	4.25
3.	10	Female	7.00-	6.00-	0.50	4.58
5.	10	Female	3.50-	5.50-	0.50	4.15
Control			5.03-	3.56-	3.84	
8.	19	Male	2.00-	2.00-	4.00	5.44
9.	18	Male	2.50-	5.50-	1.50	2.85
10.	21	Male	2.50-	5.50-	2.00	2.93
Control			5.33-	5.30-	1.27	



C<sub>1</sub>: Control for 10-year-old patients, C<sub>2</sub>: Control for 18-year-old patients.

Figure 5. Somatochart for boy patients.



C<sub>1</sub>: Control for 5-year-old patients, C<sub>2</sub>: Control for 10-year-old patients.

Figure 6. Somatochart for girl patients.

scribed. McKusich number is 185300. Frequent. Our case is a 18-year-old boy. His extremities and widths are proportionally lower than that of the control (Fig. 4).

*Patient 9 and 10. (Cockayne syndrome)*

Loss of adipose tissue is characteristic to this syndrome from early infantile period. Craniofacial dysmorphism. Rapid ageing. Microcephaly, expressed dorsal kyphosis, big hands and feet.

Autosomal recessive inheritance. Locus is not known, McKusick number is 216400. Frequency is not known.

Our patients are two elder brothers from a four-sibling family. They have a younger sister and a brother, none of them are affected. Our younger patient (9) is 18, the older one (10) is 21 years old. Their proportions are similar: the extremities and biacromial and iliocrystal widths and most of the skinfolds are lower than that of the controls but it is remarkable that the humerus and femur widths are proportionally larger. This is probably in connection with the rapid ageing, which is one of the obligatory signs of this syndrome.

The somatotype components of the patients are shown in Table 1. They were grouped according to their age and sex. The components of the corresponding control groups are also shown. The individual somatoplots are compared with the control for the boys on the Figure 5 and for the girls on Figure 6. It is remarkable that the highest values of the SAD – therefore the greatest distances from the control – are in the cases with mucopolysaccharidosis. It is also interesting that the proportionality profiles of these patients are similar to each other but differ from other cases. Great distance from the control somatoplots in the Sturge-Weber case was also found.

Our intention was to call the attention to the importance of this kind of examinations. The presented cases are not enough to give correct conclusions on the mentioned gene effect on the body shape. It is important to collect the similar cases.

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