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RETARDATION OF SOMATIC DEVELOPMENT IN DOWN'S DISEASE (MONGOLISM)*

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Anthropometric data obtained in 80 patients with Down's disease are presented. In some cases obstacles of a technical nature have made it impossible to take all the measurements planned for the purposes of this study. This explains why in the tables the number of patients varies between 70 and 80.

Horváth [8] summarized the obligate and facultative characteristics of Down's disease (mongolism) as follows.

I. Obligate characteristics:

(1) Obliquely set eyes; (2) oligophrenia; (3) microcephaly; (4) muscular hypotony; (5) a disproportionately built body.

II. Facultative characteristics:

(1) Brachycephaly; (2) epicanthus; (3) fissured lips; (4) scrotal fissuration of tongue; (5) gothic palate; (6) anomalous dental development; (7) raucous, lowpitched voice; (8) clinodactyly; (9) wide gap between big toe and second toe; (10) rectus diastasis; (11) arthrochalasis; (12) hypogenitalism; (13) marmorated skin; (14) acromicria.

Some of these characteristics are related to the progressive somatic developmental retardation, to which attention has been directed by many earlier authors [4, 10, 12, 14, 17, 19]. Several of them have studied growth, weight and head circumference in Down's disease [1, 6, 11, 14, 17, 18]. On other hand, it is remarkable that the literature available to us, although it embodies practically all the work done in the last twenty years, should contain no comprehensive account of detailed anthropometric investigations. This is a point worthy of attention, for while in the light of present-day knowledge many of the somatic symptoms in Down's disease find their explanation in functional disturbances of the neuroendocrine system [5, 9, 19], it is anthropometry which, *e.g.* by revealing the disharmonious nature of ossification processes — might clear the diagnosis of such disturbances, and thus bring us nearer to the pathology of Down's disease.

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Methods

Body weight, height and head circumference were measured. In addition, BREITMANN's [3] method was followed. The essence of his method is that instead of characterizing the individual parts of the body by absolute measurements, it gives their measurements in per cents of standing height. The "normal" average values calculated by BREITMANN [3] from his mean values for both sexes referred to the adult age group from 20 to 40 years. The average age of our case material was 10 years (range: 4 to 18 years; for scatter, our text-figures are self-explanatory). Consequently, the data obtained by us do not bear direct comparison with those of BREITMANN [3]. If here and there we nevertheless do compare them, we do so merely to provide an additional basis for the evaluation of our own figures.

BREITMANN's [3] system involves the following measurements:

A) In length:

I Upper face: from the highest point of the frontal bone to the tip of the nose.
II Lower face: from the tip of the nose to the lowest point of the chin.
III From the lower border of the chin to the jugular incisure.
IV From the jugulum to the middle of the intermammillary line.
V From the middle of the intermammillary line to the navel.
VI From the navel to the middle of the line between the left and right inguinal regions.
VII The line from the middle of the inguinal region to the inferior pole of the patella.
VIII Lower leg: from the inferior pole of the patella to the highest protruding point of the medial malleole.
IX Height of foot: from the floor to the middle of the line between the medial malleoli.

B) In width:

X Half the interacromial distance.
XI Half the intermammillary distance.
XII Length of foot: from the heel to the tip of the big toe.
XIII From the acromial joint to the middle of the elbow.
XIV From the elbow to the radiometacarpal joint.
XV From the radiometacarpal joint to the tip of the middle toe.

From the relative measurements mean values were calculated. The standard deviation was computed by the formula

$$S = \sqrt{\frac{\Sigma(x - \bar{x})^2}{n - 1}}$$

All measurements were made on the left half of the body, with the use of a MARTIN-type anthropometer. The following additional measurements were taken:

1. Chest circumference (during expiration).
2. Sagittal diameter of chest.
3. Transversal diameter of chest.
4. Head length.
5. Head width.
6. Morphological face length.
7. Distance between zygomatic arches.
8. Width of nose.
9. Length of nose.

From these additional measurements the following indexes were calculated:

1. $\text{Thoracic index} = \dfrac{\text{Sagittal diameter of chest} \times 100}{\text{Transverse diameter of chest}}$

2. $\text{Relative head circumference} = \dfrac{\text{Head circumference} \times 100}{\text{Standing height}}$

3. $\text{Cephalic index} = \dfrac{\text{Head width} \times 100}{\text{Head length}}$

4. Morphological face index $= \frac{\text{Morphological face length} \times 100}{\text{Distance between zygomatic arches}}$

5. Relative chest circumference $= \frac{\text{Chest circumference} \times 100}{\text{Standing height}}$

6. Nasal index $= \frac{\text{Width of nose} \times 100}{\text{Length of nose}}$

Finally, the question was studied whether the rule of STRATZ — the validity of which has been questioned — could be applied to children with Down's disease; this involved the determination of the proportion of head length to body length in the various age groups. In index studies, tape measures, cranial and pelvic calipers were used.

Results and discussion

Our results are presented in graphs and tables.

Fig. 1 gives the height measurements of 73 children with Down's disease and compares them with the normal range as published in the pocket manual of the Hungarian medical superintendents of schools [13]. In the series, Down's disease growth retardation was found to increase with advancing age; only about 13 per cent had reached or exceeded the height normal for their age.

Fig. 2 compares the body weight of 62 patients with the normal average weights in [13]. Although there is occasional weight deficiency, dispersion is much wider with this measurement than with height. A considerable proportion of the patients attained or exceeded the weight normal for their age.

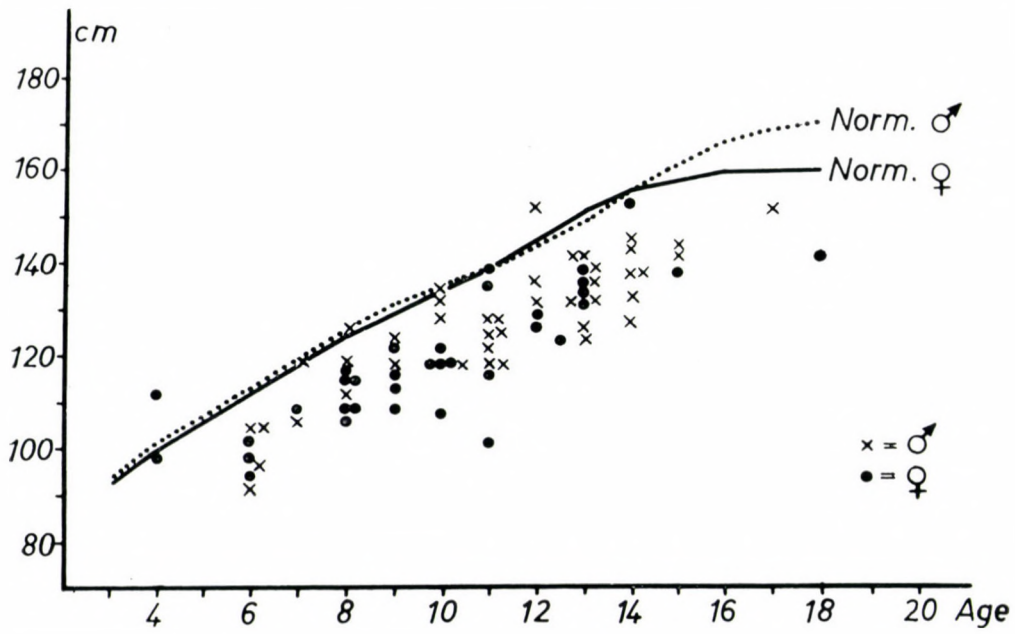

Fig. 1. Height of 77 children with Down's disease

Comparison of Figs. 1 and 2 shows that moderate dwarfism and a retardation of weight may be characteristic of Down's disease in individual cases, but are not necessarily associated with the condition.

Fig. 2b presents the body weight of 49 patients in relation to body length. In 37 cases it was found to exceed body weight corresponding to body length, and in 12 cases it was less by an average of 1.5 kg.

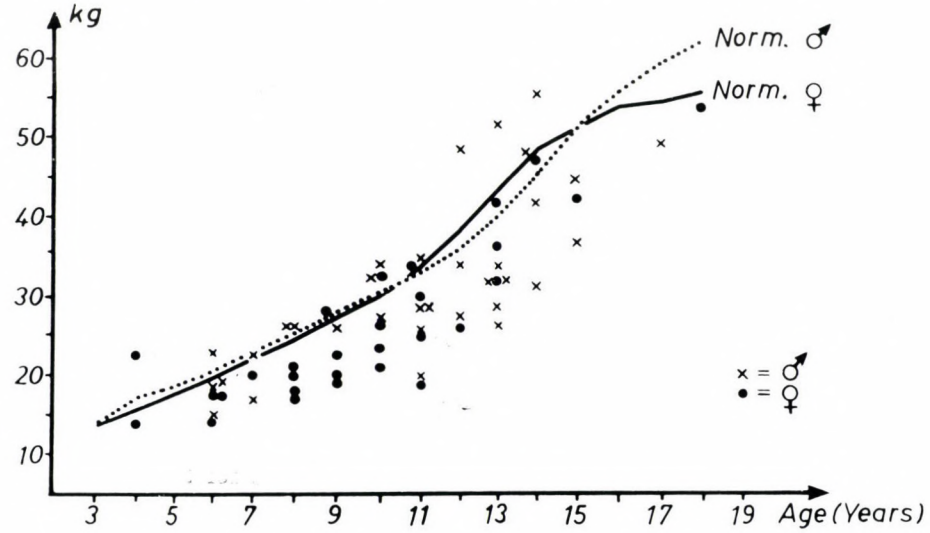

Fig. 2. Changes in body weight of 62 patients compared with mean values normal for age

Table 1a gives the body weight in relation to body length of 13 patients not included in Fig. 2b. It reveals that, although this weight may fall rather short of what is normal for age, it frequently exceeds it by 10 kg. and exceptionally by as much as 20 kg.

On comparing the data in Figs. 2 and 3 as well as in Table 1a one finds that in 12 patients out of 62 the weight deficiency was real, and in 35 out of 62 patients only apparent; 24.2 per cent were overweight in relation to the average weight for their age. These findings are, in some respects, in agreement with those of earlier authors. In the series of Roboz [17], comprising 145 cases, 20 per cent were underweight, with 80 per cent attaining or exceeding the average weight normal for body length.

Benda [1] states that the birth weight of patients with Down's disease is lower than normal while their body length is normal. Øster [14], studying a series of 65 newborns, found that their average weight was 2774 g. and their average body length 48.1 cm.

Table 1a

Body weight of 13 patients in relation to age and height

Case No.	Initials	Sex	Age	Height (cm.)	Body weight (kg.)	In relation to			
						age		height	
						+kg.	−kg.	+kg.	−kg.
1.	E. M.	♀	10	117.8	32.5	2.20	—	10.50	—
2.	F. A.	♀	13	137.5	41.5	—	2.20	9.60	—
3.	L. Á.	♀	18	141.0	54.0	—	1.80	20.00	—
4.	S. J.	♂	14	144.0	55.5	9.90	—	19.20	—
5.	V. O.	♂	15	143.5	45.0	—	6.30	9.00	—
6.	K. I.	♂	12	153.3	49.0	12.70	—	6.50	—
7.	S. J.	♂	13	124.5	32.0	—	8.40	7.00	—
8.	L. P.	♂	11	128.5	34.4	1.40	—	7.40	—
9.	Z. L.	♂	6	97.0	23.0	2.90	—	8.20	—
10.	B. J.	♂	13	142.3	51.7	11.30	—	16.70	—
11.	B. P.	♂	14	138.5	48.0	2.40	—	16.00	
12.	Z. J.	♂	14	145.0	42.0	—	3.60	5.50	—
13.	F. J.	♂	17	151.0	49.5	—	9.90	8.50	—

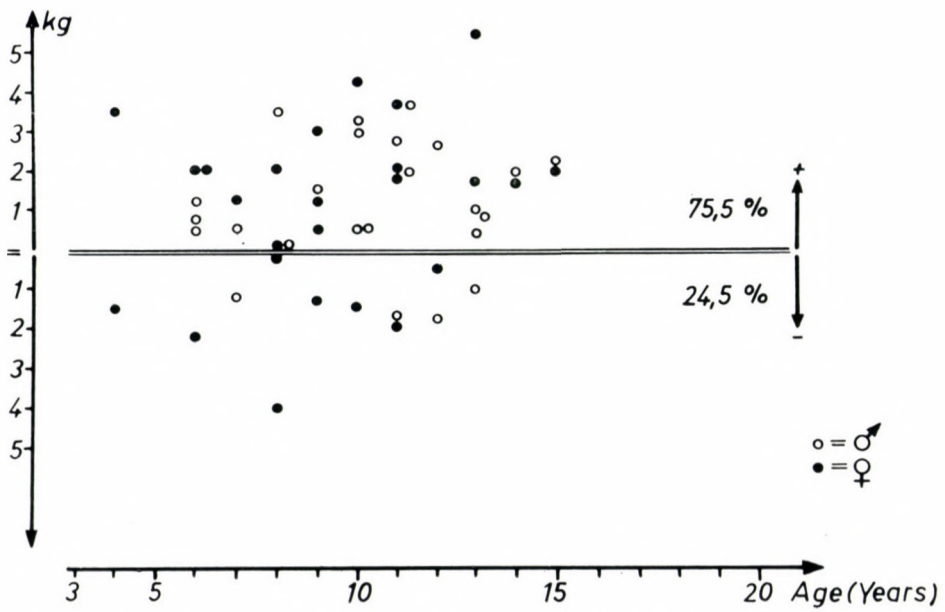

Fig. 2b. Body weight of 49 patients in relation to body length. The double horizontal line indicates body weight corresponding to body lenght, and the dots show the degree of deviation, for each case in kg. in relation to body weigth corresponding to body length. • = girl patients, o = boy patients

In Fig. 3 the head circumference of 77 patients is compared with the means for normals established by SZONDI [19] a long time ago and no longer generally accepted. They are used here on the ground that they refer to material living in similar peristatis. The data in Fig. 3 point to marked microcephaly. The head circumference of only one boy and one girl attained, respectively exceeded, the level normal for their age. However, as absolute head circumference is not suitable for deciding whether microcephaly is real or apparent, Fig. 4 presents the relative head circumferences of our patients. In relation to SZONDI's normal values for relative head circumferences corresponding to body length, our material shows very considerable deviation. In 69 per cent the microcephaly was real. But in 31 per cent it was only apparent; that is, absolute head circumference reached, or even exceeded, that corresponding to actual body length, but not to body length normal for age.

It merits mentioning that JØRGENSEN et al. [11] found the external cranial volume to be the same in normal children and children with Down's disease.

Tables 1b, 2 and 3 present the body proportions of 77 patients according to BREITMANN [3]. On comparing them with BREITMANN's normal values we find that a disproportionately built body is a definite characteristic of Down's disease. Considerable differences are demonstrable in measurements of the head (Breitmann I and II) and the leg (Breitmann VII and VIII).

Table 1b

Body proportions of boy patients according to Breitmann

Measurement	Normal	Down's disease	S ±
I.	8.85	10.00	1.61
II.	4.21	5.20	0.84
III.	5.79	4.80	1.35
IV.	6.84	7.80	0.92
V.	13.66	14.40	1.69
VI.	10.00	10.00	1.94
VII.	26.14	21.40	1.99
VIII.	20.33	21.90	1.93
IX.	4.21	4.50	0.85
X.	9.50	10.60	1.53
XI.	6.33	5.10	0.64
XII.	14.50	15.00	0.93
XIII.	18.00	18.00	2.00
XIV.	14.50	14.70	1.30
XV.	10.50	10.40	1.59

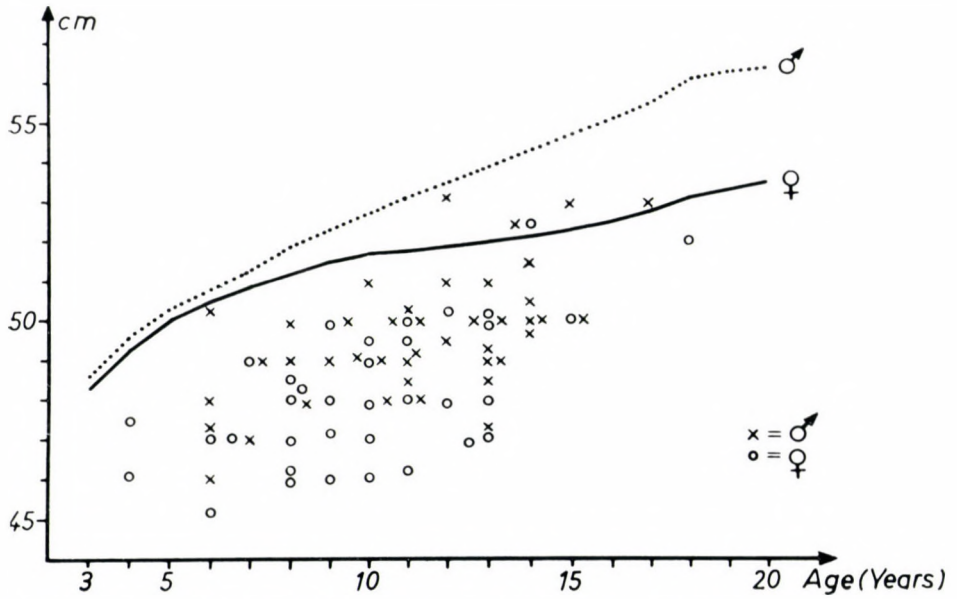

Fig. 3. Changes in absolute head circumference of 77 patients. Solid lines indicate normal means. ○ = girl patients, × = boy patients. The data point to marked microcephaly

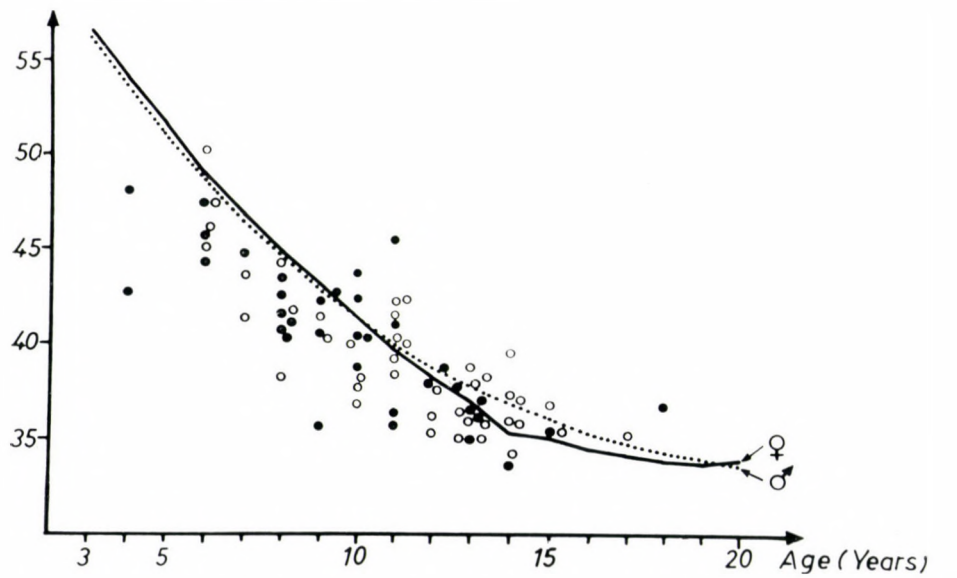

Fig. 4. Changes in relative head circumference of 77 patients. Solid lines indicate normal means. On the vertical axis figure the values for relative head circumference. ○ = girl patients, ● = boy patients. Microcephaly is real in most cases, and apparent in the rest

Table 2

Body proportions of girl patients according to Breitmann

Measurement	Normal	Down's disease	S ±
I.	8.85	10.31	2.13
II.	4.21	5.33	0.97
III.	5.79	5.33	1.60
IV.	6.84	8.28	1.20
V.	13.66	14.69	1.70
VI.	10.00	10.80	1.79
VII.	26.14	19.56	2.13
VIII.	20.33	21.65	2.22
IX.	4.21	4.27	0.68
X.	9.50	10.86	0.95
XI.	6.33	5.13	0.65
XII.	14.50	15.15	1.21
XIII.	18.00	17.68	2.38
XIV.	14.50	14.14	1.73
XV.	10.50	10.27	1.25

Table 3

Body proportions of patients according to Breitmann ; average means for both sexes

Measurement	Normal (boy + girl)	Down's disease (boy + girl)	S ± (boy + girl)
I.	8.85	10.15	1.87
II.	4.21	5.25	0.90
III.	5.79	5.06	1.47
IV.	6.84	8.04	1.01
V.	13.66	14.55	1.69
VI.	10.00	10.40	1.86
VII.	26.14	20.48	2.06
VIII.	20.33	21.77	2.07
IX.	4.21	4.38	0.76
X.	9.50	10.73	1.24
XI.	6.33	5.11	0.64
XII.	14.50	15.07	1.07
XIII.	18.00	17.84	2.19
XIV.	14.50	14.42	1.51
XV.	10.50	10.33	1.42

Fig. 5 is a diagrammatic illustration of the disproportionate measures in length in 77 patients and of the normal averages.

As regards the Breitmann VII measurement, which essentially refers to the length of the femur, we find considerable growth deficiency in patients with Down's disease. Several authors have reported disturbances in ossifi-

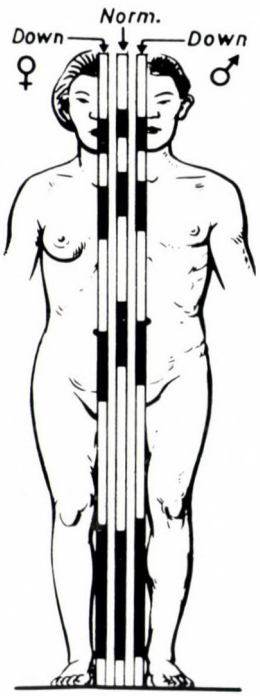

Fig. 5. Means of longitudinal body proportions according to BREITMANN, represented diagrammatically. Note characteristic deviations in head and leg measurements

cation. INGALLS [10] points to the characteristic shortness of the extremities and the growth deficiency of the long bones. He mentions that HEFKE found shortness of the metacarpal bones in 62 per cent of his series. BENDA [1] states that in Down's disease the neck of the femur is maldeveloped, and its head is poor in calcium. RANSCHBURG [16] thinks ossification is normal in Down's disease.

In normal children the lower extremities grow at a higher rate than do the trunk and the other parts of the body. At birth the relative length of the leg is about 40, but in adolescence it gradually grows to about 51, *i.e.* the value for adults [14]. In the tables annexed the relative length of the leg is the sum of the values for measurements VII, VIII and IX. For our series this is 46.63, or not much more than the normal for the 5-year-old child.

The changes in the relative length of the leg are shown in Fig. 6. They are due primarily to deficiency in the growth of the femur.

Fig. 7 demonstrates that in only one of our patients did the length of the thigh in relation to body length reach BREITMANN's value of 26 per cent.

In Down's disease, the proportions of the upper body, especially of the head, likewise show characteristic deviations from the normal averages. Whereas in the normal case the sum of the Breitmann I—V values is 39.35, in mongoloids it is an average 43.05. The relative head length (Breitmann I + II) is in the normal case 13.06, and in Down's disease an average 15.40. These data tend to prove that the Stratz numbers expressing the normal ratio of head length to body length should not be related to patients with Down's disease.

Figs. 8 and 9 present the values for relative head length in 36 girls and 43 boys with mongolism, and bring them into relation with the Stratz norms for the various age groups. On the evidence of these two figures, relative head length exceeds in the majority of the cases the head length corresponding to age (in 60.3 per cent of the boys and 57.3 per cent of the girls). INGALLS [10] refers to BULLARD who studied the skull of 25 patients roentgenologically, and found that its narrowness was characteristic not of microcephals but brachycephals, the transverse diameter being rather wide in relation to the sagittal diameter. In BUDAY's [4] view, brachycephaly and microcephaly are both definite characteristics. Other authors, as WAGNER [20] and FATTOVICH [6], write only of brachycephaly. INGALLS [9] regards brachycephaly as an obligate symptom of 100 per cent incidence.

The cephalic index was worked out for 48 of our patients. Fig. 10 shows that 39 per cent had to be classified as hyperbrachycephalic or ultrabrachycephalic, and that isocephals and hyperisocephals too occurred. Nevertheless, brachycephaly cannot be declared an obligate symptom of mongolism since 14 per cent of the skulls studied by us came to fall in the mesocephalic zone, and 23 per cent showed different degrees of dolichocephaly.

In view of this considerable scatter, we present Fig. 11, in which the morphological face index of 47 patients is compared with BUDAY's [4] classification. It shows that 59.58 per cent fell in the category of the short-faced, and almost 20 per cent in that of the medium or long and narrow-faced individuals. Like those in Fig. 10, these data do not favour the contention that brachycephaly is always present in Down's disease.

Fig. 12 shows the relative chest circumferences, which inform about growth in both length and width.

In a considerable proportion of our cases the relative chest circumference was found to exceed the value corresponding to age. This, however, cannot mean more than that in our material there was a lack in height in relation to age. The "pudgy" figure of the body, believed to be characteristic of mongolism,

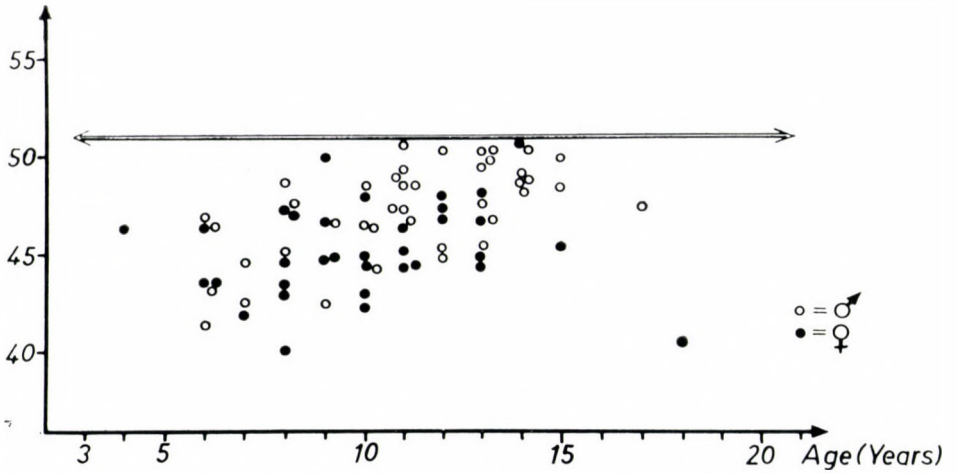

Fig. 6. Changes in the relative length of the leg in 75 patients. Double horizontal line indicates average value. On the vertical axis figure the index numbers (sums of Breitmann VII, VIII and IX measurements) of the relative length of the leg. ● = girl patients, ○ = boy patients

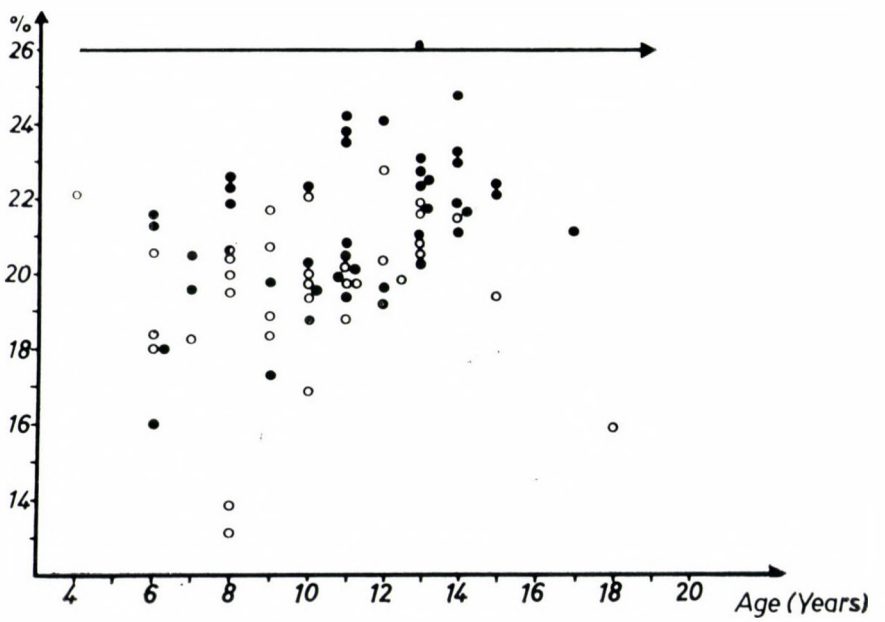

Fig. 7. Changes in relative length of femur in 77 patients. Horizontal line indicates average level according to BREITMANN in percentage related to body length. ○ = girl patients, ● = boy patients

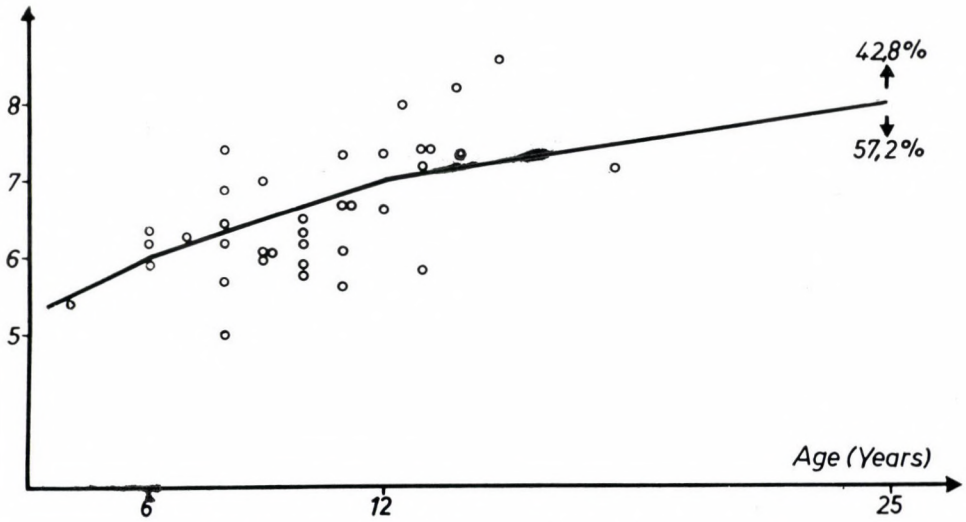

Fig. 8. Changes in the Stratz number in 36 girl patients

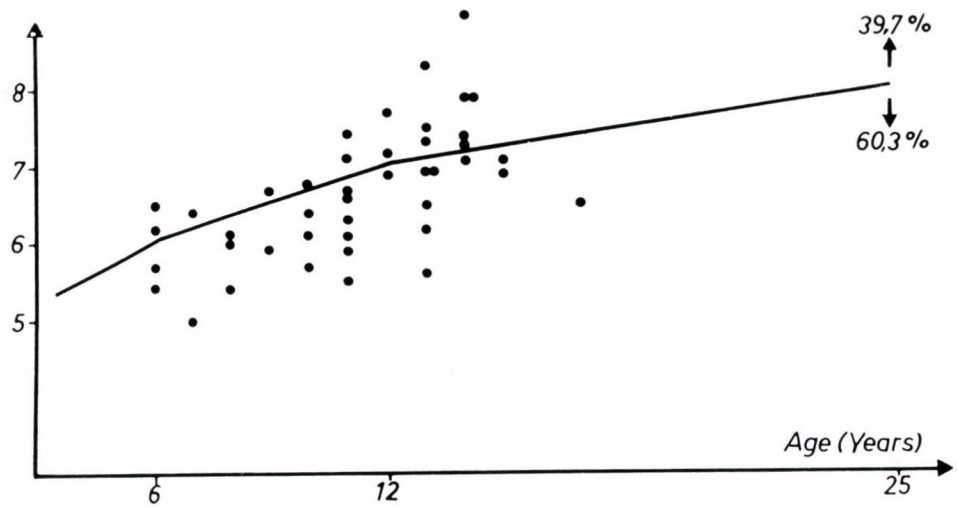

Fig. 9. Changes in the Stratz number in 43 boy patients

can in no way follow from the high values obtained for relative chest circumference, since pudginess presupposes, among other things, a wide chest. Following BRUGSCH' classification, Fig. 13 shows that — on the basis of their relative chest circumference — 63 of our 78 cases belonged in the category of medium or narrow-chested individuals.

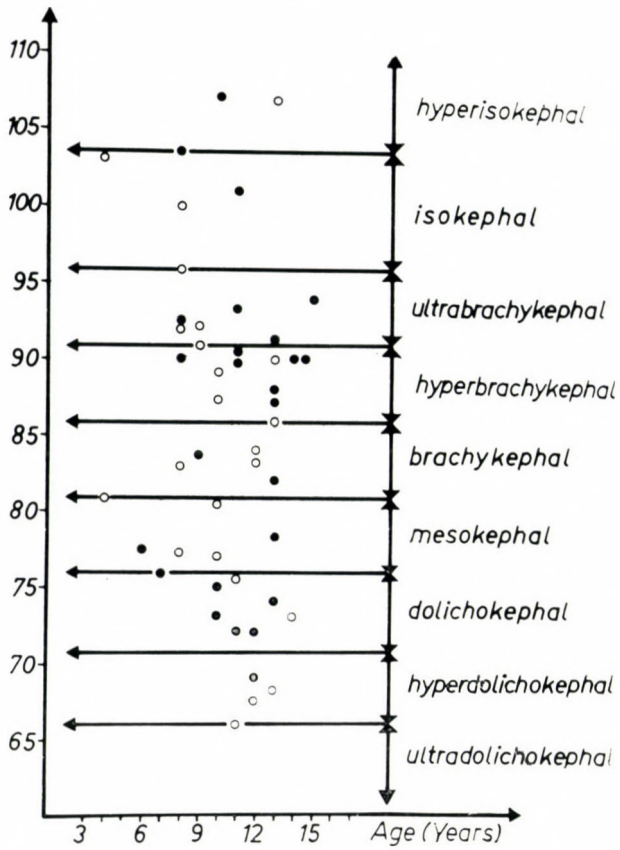

Fig. 10. Changes in the cephalic index in 48 patients. The index values show very wide scatter over the various zones; brachycephaly is not characteristic. ○ = girl patients, ● = boy patients

The average thoracic index was 75.13 for girls, 72.73 for boys, and 73.93 for both sexes. The range was from 62.22 to 86.10 for boys, and from 66.67 to 84.70 for girls.

The mean nasal index values were: 72.79 for boys, 75.01 for girls, and 73.80 for both sexes. The extremes were 57.10 and 90.90 in the case of boys, and 62.90 and 100 in that of girls. As reflected in these means, the majority of our patients were cammaerrhines, the rest leptorrhines and mesorrhines.

According to BUDAY [4], in mongoloids the retardation in physical development becomes less and less apparent after the 15th year of life. The findings

in our five patients over 15 years of age contradict this view. Small as their number is, they support the experience that the truly distinctive physical characteristics of the condition become increasingly conspicuous as time goes on. The same conclusion can be drawn from the empirical fact that it is exceedingly difficult to diagnose Down's disease at birth and in early infancy; in ØESTER's

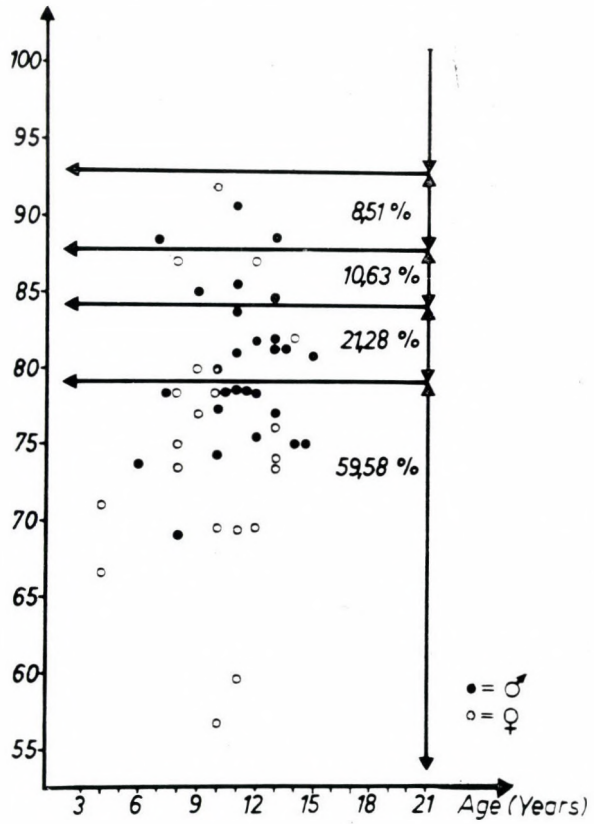

Fig. 11. **Changes** in the morphological facial index. By BUDAY's classification 8.51% of the **cases had long** and narrow, 10.63% medium-sized, 21.28% short and broad faces, and 59,58% **fell into** the very short-faced category. ○ = girl patients, ● = boy patients

[14] **opinion** the diagnosis must be left open until some time later. The expe**rience of JULESZ** [12] is that the syndrome is seldom recognized before the **patient is** at least six months old.

Not included in the annexed Tables are the data obtained in patient K. L., **a** male aged 35. His longitudinal body proportions according to Breit**mann are** presented in Fig. 14.

This case, too, is conspicuous for a high value for relative head length **and a small** one for the relative length of the leg. As in our other patients,

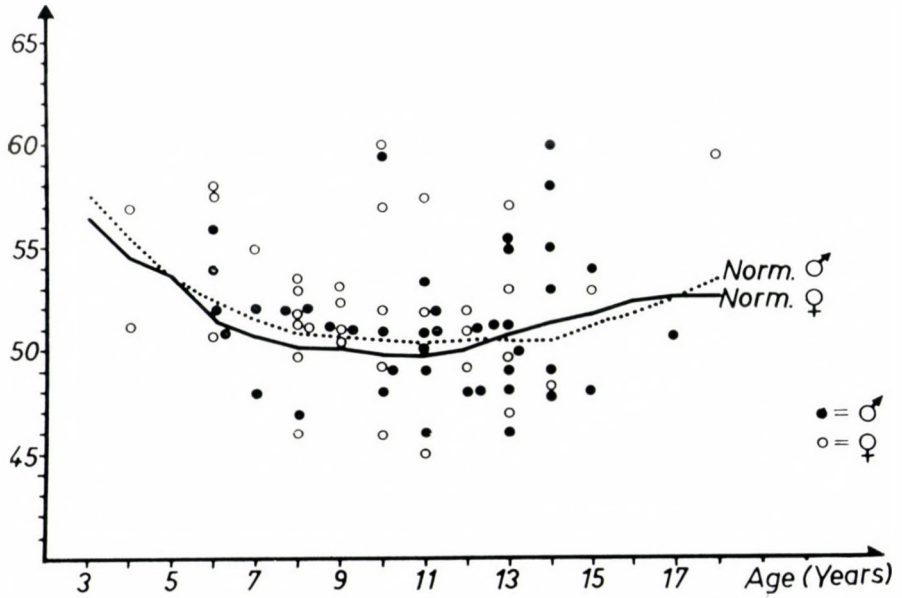

Fig. 12. Relative chest circumference values. Solid line = normal girls; dotted line = normal boys; ○ = girl patients; ● = boy patients

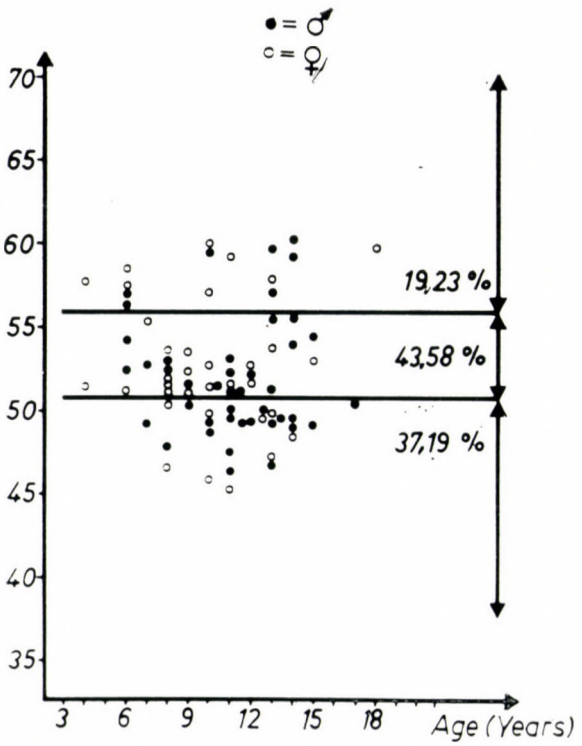

Fig. 13. Changes in relative chest circumference based on BRUGSCH' classification; in 43.58% medium-sized, and in 37.19% narrow. ○ = girl patients, ● = boy patients

these values produce characteristic deviations. Of the anthropometric index figures of K. L. the following are characteristic.

1. Relative chest circumference is 44.6, which means that the patient is narrow-chested. This value is so low that it does not even appear among the average values for normals. It should be noted that the patient's height is 150 cm., corresponding to that of a child of 13 years, and his chest circumference 67 cm., equalling that of a child of 12. Thoracic index is 64.1.

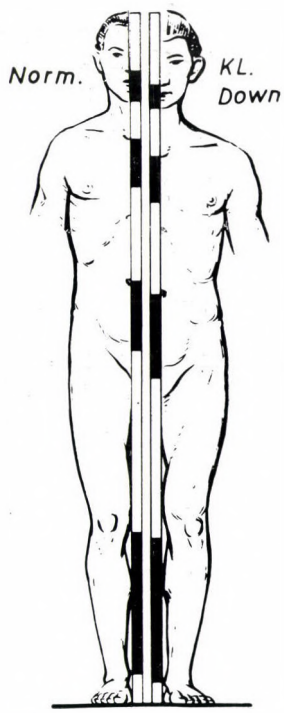

Fig. 14. K. L., male aged 35; changes in longitudinal body proportions according to BREITMANN. Normal means on left side of the figure

2. Head circumference is 50 cm., which corresponds to an age of 6 years, and so indicates marked microcephaly. Relative head circumference is 33.3, meaning that the microcephaly is real. Cephalic index is 90.1, indicating hyperbrachycephaly.

3. Morphological facial index is 62.2, showing that the face is extremely short.

A similar case has been described by FATTOVICH [6]. He kept his patient under observation from the age of 6 to his death at 32 years of age. Body height was 98 cm, and body weight 15 kg., at the age of 6 years. At 22 years the corresponding measurements were 140 cm. and 42 kg. There was marked brachycephaly with flat occiput, gothic palate, etc. FATTOVICH observed the distinctive characteristics of Down's disease to grow more conspicuous with advancing age.

Summary

Eighty patients, 36 girls and 44 boys with Down's disease have been studied anthropometrically. The average age was 10 years.

1. A disproportionately built body is characteristic of the condition. It is due to growth deficiency in the relative length of the lower leg and to shortness of the femur.

2. The rule of STRATZ cannot be applied in Down's disease, for relative head length in most cases exceeds that corresponding to age.

3. Consequently, brachycephaly is not obligate symptom. In our series it was absent in 37 per cent, and 23 per cent revealed varying degrees of dolichocephaly.

4. Microcephaly is an obligate symptom. It may be real or apparent. In our series it was real in 69 per cent and apparent in 31 per cent.

5. Retardation in height and moderate dwarfism, but not weight deficiency, are characteristic in most cases of Down's disease. In our series 24.2 per cent were overweight in relation to age.

The obligate characteristics of Down's disease become increasingly apparent in time, i.e. somatic development is characterized by progressive retardation.

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BEITRÄGE ZUR VERZÖGERUNG DER SOMATISCHEN ENTWICKLUNG BEI DER DOWNSCHEN KRANKHEIT

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Einige wichtigen Körpermaße wurden an 80 Downschen Kranken mit der üblichen, Breitmannschen anthropometrischen Methode untersucht. Im Krankengut befanden sich 36 Mädchen und 44 Knaben. Das Durchschnittsalter betrug 10 Jahre. Aus den Untersuchungsergebnissen lassen sich folgende Schlüsse ziehen:

1. Für die Downsche Krankheit ist der dysproportionierte Körperbau kennzeichnend, was in erster Linie der geringeren relativen Länge des Femurs zugeschrieben werden kann.

2. Auf die Downsche Krankheit bezieht sich das Stratzsche Gesetz nicht. Die relative Kopfhöhe übersteigt die dem Alter entsprechende relative Kopfhöhe.

3. Die Brachyzephalie ist kein obligates Kennzeichen der Downschen Krankheit. In 37% der Fälle kann sie nicht nachgewiesen werden, in 23% zeigten diese Kranken sogar eine Dolichozephalie verschiedenen Grades.

4. Die Mikrozephalie ist hingegen ein obligates Symptom. In 69% der Fälle wurde eine echte, in 31% eine scheinbare Mikrozephalie beobachtet.

5. Das geringere Körpergewicht ist kein typisches Kennzeichen. In 24% der Fälle war das Gewicht der Kranken höher als das dem Alter entsprechende Gewicht.

6. Die somatische Entwicklung der Downschen Kranken wird durch progressive Verzögerung gekennzeichnet. Das konstitutionale Bild wird mit zunehmendem Alter immer ausgeprägter.

ОТ ОТСТАВАНИИ СОМАТИЧЕСКОГО РАЗВИТИЯ У БОЛЬНЫХ БОЛЕЗНЬЮ ДАУНА

В. ГЁЛЛЕС

Автор обычным антропометрическим методом Брейтманна исследовал несколько важных размероов тела у 80 больных, страдающих болезнью Дауна. В изученном материале больных было 36 девочек и 44 мальчика. Средний возраст больных составлял 10 лет. Из результатов исследований можно делать следующие заключения:

1. Для болезни Дауна характерна диспропорция телосложения, что в первую очередь является результатом отставания относительной длины бедренной кости.

2. К лицам, страдающим болезнью Дауна, не относится правило Штратца. В исследованных автором случаях относительная высота головы превышает высоту головы, соответствующую возрасту.

3. Брахицефалия не является обязательным симптомом болезни Дауна. В 37% случаев не удавалось выявить брахицефалию, а в 23% случаев даже обнаруживалась долихоцефалия различной степени.

4. Обязательным симптомом болезни Дауна является микроцефалия. В 69% случаев автор обнаруживал истинную, а в 31% — кажущуюся микроцефалию.

5. Отставание в весе тела не является характерным симптомом болезни Дауна. В 24,2% случаев вес тела превышал средний вес соответствующего возраста.

6. Соматическое развитие больных, страдающих болезнью Дауна, характеризуется прогрессивным замедлением. Картина конституции по мере повышения возраста становится все более выраженной.

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