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Intrauterine Growth in Autosomal Trisomy Syndromes

By

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During the period 1969 to 1972, altogether 23 newborn infants with different types of autosomal trisomy syndrome were observed. Intrauterine growth of the 11 neonates with trisomy 21 (Down's syndrome) was normal and their gestational age near term (36.3 ± 0.9 weeks). The infants with trisomy 13 (Patau's syndrome) were prematurely born (gestational age, 34.6 ± 1.2 weeks) and had a moderate but significant (−15.7 ± 6.3%) retardation in birth weight, whereas birth length was within normal limits of the corresponding gestational age. The 4 newborns with trisomy 18 (Edwards's syndrome) were mature as to gestational age (39.8 ± 1.6 weeks), but heavily retarded in both birth weight and length (−38.2 ± 4.8 and −17.6 ± 2.6%, respectively). A case of cat-eye syndrome is also presented.

The effect of chromosomal abnormalities on foetal development and birth weight has gained increasing interest. The results of several studies suggest that the newborns with chromosomal aberrations are usually small for gestational age [1]. However, the data about gestational age, placenta, birth weight, and especially the length of such neonates, are still too scarce for drawing general conclusions. This is why we report on our observations on newborns with autosomal trisomy syndromes.

Material and Methods

In the four-year period 1969 to 1972, altogether 23 newborns with autosomal trisomy syndromes, in whom gestational age, birth weight, and length were reliably known, have been observed. Out of the 23 babies, 11 displayed typical Down's syndrome with regular trisomy 21, and 4 neonates the symptoms of Edwards's syndrome with a 47.18+ karyotype. One of the 7 newborns with clinically typical Patau's syndrome had a 46,XY,D-,t (Dq/Dq)+ translocation trisomy, the karyotype of the other six was 47,D+. In addition, one case of cat-eye syndrome was observed.

Gestational age was calculated from the first day of the last normal menstrual period. For assessing intrauterine growth, a chart of our own construction [2] was used. In addition to the percentile position, in each case the percentage deviation from the mean birth weight for the relevant gestational age was estimated and plotted against the deviation of crown-heel length, obtained similarly.

Results

Results are summarized in Table I. Mean gestational age was 36.3 ±

TABLE I

Data of newborns with different types of autosomal trisomy, mean ± SE

Type of trisomy syndrome	Number of cases	Gestational age, weeks	Birth weight, g	Birth length, cm	Weight deviation, per cent	Length deviation, per cent	Placental weight, g
Down's	11	36.3 ± 0.9	2590 ± 167	48,4 ± 1.2	− 2.4 ± 3.1	− 3.6 ± 1.5	452 ± 34
Patau's	7	34.6 ± 1.2	1910 ± 224	46.0 ± 4.0	−15.7 ± 6.3	− 6.4 ± 3.8	404 ± 62
Edwards's	4	39.8 ± 1.6	1940 ± 178	44.5 ± 1.8	−38.2 ± 4.8	−17.6 ± 2.6	363 ± 70
Cat-eye	1	41	2640	50.0	−21.5	− 7.4	470

± 0.9 weeks in Down's syndrome, 34.6 ± 1.2 weeks in Patau's syndrome, and 39.8 ± 1.6 weeks in Edwards's syndrome. Only the difference in gestational age between Patau's and Edwards's syndromes proved to be significant ($p < 0.05$).

In our cases of Down's syndrome, mean birth weight was 2580 ± 167 g, slightly lower than the normal mean of the corresponding gestational age [2] and the mean deviation was also negligible (−2.4 ± 3.1%). Mean birth weight in Patau's syndrome (1910 ± ± 224 g) was 15.7 ± 6.3%, less than expected. Mean weight in Edwards's syndrome was 1940 ± 178 g, markedly lower than that expected for the respective gestational age, which represents a deviation of −38.2 ± ± 4.8 per cent. The difference in mean birth weight between Down's syndrome on the one hand and Patau's and Edwards's syndromes on the other was significant at the 5% level.

Crown-heel length showed practically no deviation from the normal mean corresponding to the gestational age, neither in our cases with Down's syndrome, nor in those with Patau's syndrome. The mean birth length of 44.5 ± 1.8 cm in Edwards's syndrome was, however, significantly different ($p < 0.01$) from the expected normal value, representing an average deviation of −17.6 ± 2.6% which also differed significantly from the mean deviations observed in both trisomy 21 and 13 (−3.6 ± 1.5%, $p < 0.001$; and −6.4 ± 3.8%, $p < 0.05$, respectively).

In Fig. 1, the individual birth weights of the newborn infants in the three groups with autosomal trisomy are plotted against gestational age on our intrauterine growth chart. It can be seen that all the birth weights of our Down cases were above the 25th percentile and therefore this group of infants did not fulfil the statistical criteria of intrauterine growth retardation. Although the birth weights of infants in the Patau group were all scattered in the lower range of normal distribution, only two of them had a birth weight lower than the 10th percentile for the respective gestational age. All the four

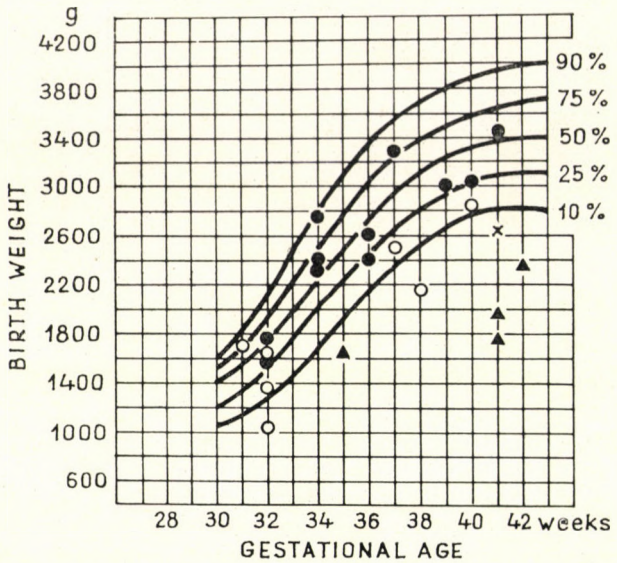

FIG. 1. Intrauterine growth in 23 newborn infants with different types of autosomal trisomy. ● Down's syndrome (n : 11); ○ Patau's syndrome (n : 7); ▲ Edwards's syndrome (n : 4); × Cat-eye syndrome (n : 1)

infants with Edwards's syndrome had a birth weight well below the 10th percentile.

Since the percentile position of birth weight in itself does not give information concerning the weight deficit in relation to body length, the percentage deviations of the two parameters have been calculated and plotted against each other in Fig. 2. It can be seen that in Down's syndrome neither birth weight nor crown-heel length showed a significant deviation from the normal. In Patau's syndrome, except one infant, a moderate weight deficit but a nearly normal length was observed. In contrast, the four infants exhibiting trisomy 18 were heavily retarded in both birth weight and crown-heel length.

Mean placental weight was 452 ± ± 34 g in Down's syndrome, 404 ± ± 62 g in Patau's syndrome, and 363 ± 70 g in Edwards's syndrome. Since certain technical errors in weighing the placentas could not be excluded, these data are not further dealt with.

Discussion

The present results confirm the observation that the different chromosomal anomalies have different effects on foetal growth. Thus, in the three classical autosomal trisomy syndromes, the birth weights in decreasing order are trisomy 21, 13, and 18.

In view of the small number of the cases, our findings may be accidental. It seems, however, interesting that our patients with Down's anomaly were not significantly retarded in intrauterine growth. A further

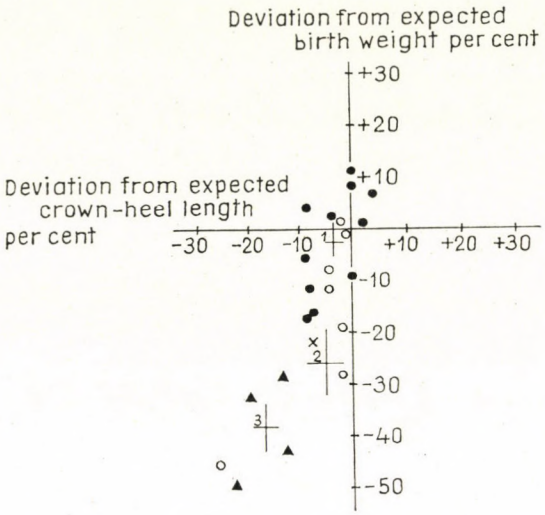

FIG. 2. Relationship between deviation from the expected weight and length in trisomic infants, expressed as a percentage of the mean value for birth weight and crown-heel length, appropriate for gestational age according to the growth standard of FEKETE et al. [2]. 1. ● Down's syndrome; 2. ○ Patau's syndrome; 3. ▲ Edwards's syndrome; 4. × Cat-eye syndrome. Mean ± SE

finding to be emphasized is that in Patau's syndrome we observed a considerably shorter gestational age and lower birth weight than those reported in previous surveys [1, 4, 6]. On the other hand, the markedly decreased growth rate of our patients with Edwards's syndrome corresponds to the data of TAYLOR [6], SCHINZEL and SCHMID [5], and LUBCHENCO [3].

Since birth length was not systematically analyzed in previous studies, the present observations might be of some interest as far as the relationship between growth rate of body weight and crown-heel length is concerned. The findings indicate that while infants with trisomy 13 were relatively long in relation to their body weight, the length and weight of the neonates with trisomy 18 were equally severely retarded. This might suggest that the former chromosomal anomaly affects foetal growth at a more advanced stage of pregnancy than the latter. Alternatively, it appears reasonable to suppose that while in Patau's syndrome growth retardation is mainly the secondary consequence of major malformations, in trisomy 18 the chromosomal anomaly may primarily impair foetal growth.

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