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A simple score to facilitate detection of congenital disorders

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A simple score has been constructed to facilitate the selection of apparently normal newborns at risk of hidden congenital disorders. Components of the score are family history of previous malformations, stillbirth or infantile death of unknown origin, intrauterine growth retardation (2 points each), and six minor malformations: antimongoloid palpebral slant, hypertelorism, preauricular fistula, simian crease, mammillary and hallucal abnormality (1 point each).

Out of 1000 consecutive neonates screened with the method 28 scored 3 or more, in 6 of whom hidden congenital abnormalities were discovered at later reexamination. At the same time only 2 hidden defects were found in the children with a neonatal score of 2 or less. Although the follow-up was incomplete, the results suggest that the score is useful in selecting infants for more complicated clinical and genetic investigations.

About 5 to 6% of live-born neonates are affected by congenital abnormalities with completely or partly genetic background [21]. Half of them show major malformations or characteristic symptoms, which makes early diagnosis easy. In about 2 to 3% of newborn infants there are, however, no obvious signs of an anomaly, and the hidden morphological or functional disorder cannot be suspected. The most effective method to prevent irreversible somatic and mental damage in such cases is the screening of unselected neonatal populations. Indeed, biochemical mass screening has saved, for example, hundreds of phenylketonuric and galactosaemic children all over the world, and serial cytogenetic investigations of unselected newborn infants are of great scientific value. In spite of this considerable progress the available biochemical and cytogenetic screening methods can detect only a limited number of abnormal conditions, and except for phenylketonuria screening, the cost of the required technical and financial investment makes these methods impracticable for mass screening purposes.

Hence, there is still need to refine the physical examination of the newborn infant. Careful observation of minor malformations offers a possibility to increase the effectiveness of early diagnosis of congenital disorders. These small morphological anomalies are in themselves clinically insignificant, but many studies suggest that they are positively correlated with significant fetal pathology [9, 23].

Marden et al. in 1964 [13] concluded that 'the detection of multiple minor anomalies may be of value alerting the physician to the existence of an obscure major abnormality or the recognition of a specific multiple anomaly syndrome'. Since then this idea has been firmly established but the principles of utilizing the minor birth defects in clinical diagnosis has not yet been put into practice.

In the last few years we have attempted to elaborate an effective but simple screening method for selected minor malformations.

The incidence of minor malformations was on the average 0.21 per neonate in full-term, well-nourished infants, and 0.76 in small-for-gestational age (SGA) newborns [16]. This high frequency of some minor defects in SGA-infants was also observed by Drillien (4), Crichton et al. [3], and Hook et al. [11].

Multiple minor malformations represent a high risk of hidden major anomalies or pathological conditions [16]. Out of 110 apparently healthy newborns with two or more minor malformations, 10 proved to have severe disorders at reexamination after the age of 1 year [14]. Since multiple minor anomalies are more common in SGA-babies, who are also known to be frequently affected by major malformations [2, 4, 14], the intrauterine malnourished newborn is considered to be at high risk for congenital abnormalities.

As to single minor malformations, several features such as a peculiar face and deformity of the ears, cannot be defined precisely, which makes the diagnosis rather subjective, as shown by a Hungarian collaborative study of 10 203 newborn infants. Some other features tend to disappear with age. For example, in a follow-up study we observed that 74% of low-set ears and 79% of high arched palate found in the newborn could not be seen after the age of 1 year [19]. These anomalies may be typical of certain syndromes, but are in themselves of little diagnostic value. In addition, some minor defects, previously regarded as important signs, are not closely correlated with any congenital disorder. This seems to be the case with the single umbilical artery [1, 6], etc. At the same time, there are some well-defined minor malformations which in themselves may indicate a certain risk of further hidden abnormalities.

Ocular hypertelorism is a characteristic symptom of at least 30 syndromes. Its recognition by inspection is often unreliable; exact measurement is necessary to avoid a false positive scoring [5, 18].

Antimongoloid (downward) slant of palpebral fissures proved to be a fairly consistent feature throughout childhood. Its distinct form occurs in 1% of neonates, and it often determines the peculiar appearance of the face and is characteristic of some special conditions.

Preauricular fistulae may be inherited in some families. They are rare in normal newborns. For example, no preauricular fistula was seen in 11 203 normal neonates. Its

appearance is often associated with other disorders.

Mammillary abnormality. Widely set, too small, absent or accessory nipples may call attention to several syndromes, and especially to disorders of the kidneys and the urinary tract [7, 8].

The *simian crease* shows a fairly close correlation with a variety of congenital anomalies. This is valid for both genetically determined [15] and environmental diseases [10].

Abnormalities of the hallux should make one to look for several malformation syndromes and for some disorders of mineral metabolism.

On the basis of these earlier observations we constructed a simple scoring system. The score is based besides six important and easily recognizable minor malformations, on intrauterine growth retardation and the family history. The first experiences with the score are presented in this study.

Material and Methods

A total of 1019 newborn infants were examined within the first 48 hours after birth. Neonates with major malformations were excluded, and only 1000 apparently normal babies were involved, irrespective of birth weight. The examinations were carried out by doctors not trained in genetics. They only received a list of the following scoring criteria.

Family history: malformation, known hereditary disease, infantile death or stillbirth of unknown origin among the siblings and parents of the index subject.

Intrauterine growth retardation: birth weight and length under the 10-percentile curve of the Hungarian standard.

Antimongoloid slant: the outer canthi fall below the line of the inner ones, as judged by the edge of a plastic ruler laid across the two inner canthi.

Hypertelorism of the eyes: the distance of the inner canthi is longer than 8% of the head circumference measured at the same level [18].

Preauricular fistula can be detected by mere inspection.

Mammillary abnormalities: accessory, hypoplastic or absent nipples, or those lying widely apart, at a distance measuring 28% or more of the chest circumference [18, 20].

Simian crease: only classic, uninterrupted lines qualified without bridge formation.

Hallucal abnormalities: broad, dorsoflected and short halluces, unusually wide gap between the 1st and 2nd toes.

Quantitation of the score was given as follows:

Positive family history	2 points
SGA-infant	2 points
Antimongoloid slant	1 point
Ocular hypertelorism	1 point
Preauricular fistula	1 point
Mammillary abnormality	1 point
Simian crease	1 point
Hallucal abnormality	1 point

All the infants who scored 3 or more were controlled at home by a district nurse, and invited to a reexamination at the age of 3 to 12 months. On this occasion a detailed paediatric examination was supplemented with cardiologic, radiologic, metabolic and karyotype investigations.

The infants with a score under 3 were observed at home, and in case of any illness were referred to our department. A detailed reexamination could, however, be carried out only in a representative sample of 58 babies with a score of 2, and 130 with a score 1 or 0.

Results

Of the 1000 newborns 747 had a normal family history, and no intrauterine malnutrition or any minor malformation. One minor anomaly occurred in 59 cases (Table I). 166 infants scored 2 points mainly because of a positive family history or intrauterine malnutrition. In 28 babies the score was 3 or more. As demonstrated by the figures, a positive family history and intrauterine growth retardation were the commonest features. The frequency of the six minor malformations examined was in accordance with data of previous studies.

The findings at reexamination are summarized in Table II. In spite of the small number of positive cases, the figures demonstrate a significantly higher frequency of hidden congenital disorders in infants scoring

Table I

Score values and distribution of symptoms in 1000 apparently normal infants at birth

	Score value						Total frequency	
	0	1	2	3	4	5	No.	Percent
Number of infants	747	59	166	17	9	2	1000	100.0
Positive family history			85	13	8	1	107	10.7
Small-for-gestational age			78	6	3	2	89	8.9
Antimongoloid palpebral slant		1	0	1	2	1	5	0.5
Hypertelorism		20	2	4	4	1	31	3.1
Preauricular fistula		0	0	0	0	1	1	0.1
Mammillary abnormality		15	1	3	4	0	23	2.3
Simian crease		22	5	4	4	1	36	3.6
Hallucal abnormality		1	0	1	0	0	2	0.2

Table II

Results of reexamination of children with different neonatal score values

Score value	No. of infants	Reexamined No.	Congenital disorder No.	
3 or more	28	27	6	
				$p < 0.01$
2	166	58	1	
1	59	28	0	
0	747	102	1	
Total	1000	215	8	

3 or more. The small number of hidden major defects in children with 2 and 1 score values suggests that a positive family history, intrauterine malnutrition or a single minor malformation alone is of little alerting value, but the combination of these should carefully be considered. A low proportion of unaffected children with a score of 0 could be reexamined. Still, all the children seen as newborns live in the district of our hospital, and none of them has been referred to us with a congenital disorder.

The reexamination revealed a congenital defect in 8 infants who at birth seemed to be normal. The following 6 infants scored 3 or more at neonatal screening.

Case 1. A boy born after 40 weeks of gestation. Birth weight, 2010 g; length, 46 cm. Both halluces were conspicuously small and dorsoflected. Score: 3. No problem had arisen until the age of 6 weeks, thereafter he failed to thrive, signs of rickets developed. Reexamination at the age of 4 months revealed a late infantile type hypophosphatasia.

Case 2. Male infant. Birth weight, 3300 g, length, 53 cm. Fifth pregnancy of the mother, the fourth resulted in stillbirth of unknown cause. The newborn had bilateral simian creases and hypertelorism. Score: 4. He was symptomless and thrived well until the age of 1 year, when a 47, XXY karyotype (Klinefelter's syndrome) was discovered.

Case 3. In a SGA male neonate (gestational age, 39 weeks; birth weight, 2100 g; length, 45 cm) antimongoloid slant of the palpebral fissures and a simian crease were noticed. Score: 4. From the fifth week of life he had been admitted several times due to recurrent pneumonias and failure to thrive. Mucoviscidosis was diagnosed and confirmed by autopsy when he died at the age of 6 months.

Case 4. Male neonate. Gestational age, 41 weeks; birth weight, 2350 g; length, 46 cm. He had antimongoloid palpebral slant, hypertelorism and a preauricular fistula on the left side. Score: 5. He was symptomless in the first weeks, but later jaundice developed. Congenital biliary aplasia was detected.

Care 5. Full-term, well-nourished male infant. He was the third child of his parents, the first died of meningocele. He had a small accessory nipple on the right side. Score: 3. At reexamination, pyuria was found. Radiologic investigation revealed bilateral dilatation of the ureters and vesicorenal reflux at the age of 7 months.

Case 6. SGA newborn girl. Gestational age, about 37 weeks. Birth weight, 1800 g; length 43 cm. She had wide set nipples (intermammillary index = 30.1%). Score: 3. She thrived well. At the age of 3 months she was referred to our department with a history of prolonged fever. Pyelonephritis and hydronephrosis on the right side were diagnosed.

Two infants with neonatal scores 2 and 0 respectively, appeared to have hidden malformations.

Case 7. SGA female newborn (gestational age, 39 weeks; birth weight,

2360 g; length, 47 cm). Score: 2. Symptomless at birth. A systolic murmur was noticed in the third week; later a ventricular septal defect was diagnosed.

Case 8. Well-nourished female neonate. Score: 0. At the age of 10 months she had prolonged fever and pyuria. X-rays revealed bilateral vesicoureteral reflux and hypoplastic left kidney.

Since this study deals with apparently healthy newborn infants, only the frequency of hidden disorders is reported. However, while screening the 1000 apparently normal neonates, 19 infants were found with various congenital diseases diagnosed in the early neonatal period. These included 2 chromosome abnormalities (Down syndrome), 1 enzymopathy (histidinaemia) and 16 gross malformations. Thus, the sum of congenital disorders was 27 in 1019 neonates, i. e. 2.65%, which is in good agreement with the international statistics excluding blood group incompatibilities and mental retardation.

Discussion

The aim of the present study was to design a simple score system to facilitate the early detection of congenital disorders in an apparently normal newborn population. A basic requirement was that unskilled observers should be able to carry out the screening quickly and without extra cost in every neonatal unit. On the basis of previous studies six minor but distinct malformations were thought to be important clues in the detection of major defects. In addition, the family history and intrauterine growth rate were chosen to contribute to the scoring system. Selection of these criteria and their relative value expressed in points was arbitrary. Previous malformations or infantile death in the family and dismaturity seemed, however, to be so strongly correlated with genetic disorders that they deserved special consideration. Combination of these phenomena with minor malformations and multiple minor malformations in themselves were also supposed to indicate a high risk for hidden major abnormalities. Babies with a score of 3 or more were regarded as high risk, and an effort was made to re-examine infants with 3 or more points. Only part of the babies scoring 2, 1 and 0 could be re-examined; the absolute number of infants followed-up in this group was, however, still 188, i.e. six times that of the children scoring 3 or more.

The results clearly suggest that newborn infants with a score of 3 or more are at high risk for hidden abnormalities. At reexamination 6 out of 27 with scores 3 or more were found to be affected by some congenital disorder, while this ratio was only 2 out of 188 among the children scoring 2 or less. Since only about 3% of all the apparently normal neonates scored 3 or more, the scoring system seems to be suitable for the selection of infants at risk.

We do not know exactly how many cases with congenital abnormalities have been overlooked, yet based on

the present results the number of such infants was probably negligible.

The individual minor malformations included in the score are by no means pathognomonic for a given group of abnormalities or syndromes. Chromosome anomalies, inborn errors of metabolism and organ malformations were all represented among the disorders discovered. The majority was clearly of genetic origin, but teratogenic abnormalities might have also been involved.

In some of the children scoring 3 or more, certain disorders may not have been recognized. Long term follow-up of such patients seems therefore justified. This is all the more warranted since in earlier studies the number of minor birth defects was positively correlated with idiopathic mental retardation [22], with hyperkinetic, aggressive and intractable behaviour [24] and even with leukaemia [12].

An advantage of the score is its simplicity. It can easily be combined with the first routine examination of the newborn, thus practically all neonates can be screened. The efficiency of the programme was 100%, the quality of the work of the different observers could be checked only indirectly, but in a previous study the frequency of clinically recognizable autosomal anomalies observed by doctors not specially trained correlated well with the data of cytogenetic screening programmes [17].

A further advantage of the screening was that it stimulated the observers to carry out a more careful physical examination of the infants. This resulted in more exact recording of major abnormalities and also helped to detect some more minor malformations not included in the score.

If our results will be confirmed by a larger prospective study, this or a similar simple score might help in selecting the newborns at risk. This would be especially beneficial in countries where facilities for mass screening employing sophisticated genetical and biochemical investigations are not available.

References

1. Bryan, E. M., Kohler, H. G.: The missing umbilical artery II. Paediatric follow-up. Arch. Dis. childh. **50,** 714 (1975).
2. Chen, A. T. L., Chan, Y.-K., Falek, A.: The effects of chromosome abnormalities on birth weight in man. II. Autosomal defects. Hum. Hered. **22,** 209 (1972).
3. Crichton, J. U., Dunn, H. G., McBurney, A. K., Robertson, M.-A. Tredger, E.: Minor congenital defects in children of low birth weight. J. Pediat. **80,** 830 (1972).
4. Drillien, C. M.: The small-for-date infant: Etiology and prognosis. Pediat. Clin. N. Amer. **17,** 9 (1970).
5. Feingold, M., Bossert, W H.: Normal values for selected physical parameters: An aid to syndrome delineation. Birth Defects Orig. Art. Ser. **10/13,** 1 (1974).
6. Froehlich, L. A., Fujikura, T.: Follow-up of infants with single umbilical artery. Pediatrics **52,** 6 (1973).
7. Goeminne, L.: Synopsis of mammorenal syndromes I. Humangenetik **14,** 170 (1972).
8. Goeminne, L.: Synopsis of mammo-renal syndromes II. Humangenetik **17,** 271 (1973).
9. Hook, E. B.: Some general considerations concerning monitoring: application to utility of minor defects as markers. In: Hook, E. B., Janerich, D. T., Porter, I. H. (Eds): Monitoring, Birth Defects and Environment. Academic Press, New York 1971, p. 177.

10. Hook, E. B,. Bonenfant, R., Powers M. L., Greenberg, M., Shapiro, L. R.: The human simian crease and its variants. Birth Defects Orig. Art. Ser. **10/7,** 7 (1974).
11. Hook, E. B., Marden, P. M., Reiss, N. P., Smith, D. W.: Some aspects of the epidemiology of human minor birth defects and morphological variants in a completely ascertained newborn population (Madison study). Teratology **13,** 47 (1976).
12. Kobayashi, N., Furukawa, T., Tkatsu, T.: Congenital anomalies in children with malignancy. Paediat. Univ. Tokyo **16,** 31 (1968).
13. Marden, P. M., Smith, D. W., McDonald, M. J. Congenital anomalies in the newborn infant, including minor variations. J. Pediat. **64,** 357 (1964).
14. Meggyesy, V., Méhes, K.: Screening der Neugeborenen. Kinderärztl. Prax. **45,** 117 (1977).
15. Méhes, K.: Simian crease. Acta paediat. Acad. Sci. hung. **13,** 335 (1972).
16. Méhes, K., Mestyán, J., Knoch, V., Vincellér, M.: Minor malformations in the neonate. Helv. paediat. Acta **28,** 477 (1973).
17. Méhes, K.: Clinically recognisable autosomal anomalies in neonates. Lancet **1,** 1262 (1973).
18. Méhes, K., Kitzvéger, E.: Inner canthal and intermamillary indices in the newborn infant. J. Pediat. **85,** 90 (1974).
19. Méhes, K.: A follow-up study of infants with minor malformations. In: Szabó, G., Papp, Z. (Eds): Medical Genetics. Akadémiai Kiadó, Budapest (in press).
20. Pelz, L.: The intermamillary index in children. Hum. Hered. **22,** 204 (1972).
21. Polani, P. E.: Incidence of developmental and other genetic abnormalities. Proc. roy. Soc. Med. **66,** 1118 (1973).
22. Smith, D. W., Bostian, K. E.: Congenital anomalies associated with idiopathic mental retardation. J. Pediat. **65,** 189 (1964).
23. Smith, D. W.: Minor malformations: their relevance and significance. In: Hook, E. B., Janerich, D. T., Porter, I. H. (Eds.): Monitoring, Birth Defects and Environment. Academic Press, New York 1971, p. 169.
24. Waldrop, M. F., Pederson, F. A., Bell, R. Q.: Minor physical anomalies and behavior in preschool children. Child Develop. **39,** 391 (1968).

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