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Congenital anomalies in black South African liveborn neonates at an urban academic hospital

S. D. Delport, A. L. Christianson, H. J. S. van den Berg, L. Wolmarans, G. S. Gericke

Study objective. The aim was to study the spectrum of clinical problems and outcomes in infants born at an urban academic hospital. In consequence, as part of the overall study, the incidence of congenital anomalies and the outcomes of affected infants were recorded.

Design. This was a prospective, hospital-based study, undertaken on liveborn infants born over a 3-year period, 1 May 1986 to 30 April 1989.

Setting. Kalafong Hospital, Pretoria.

Main results. A total of 17 351 liveborn infants was examined and the total congenital anomalies incidence was 11,87 per 1 000 livebirths. The central nervous system was the system most frequently involved (2,30 per 1 000 livebirths), followed by the muscuioskeletal system (2,13 per 1 000 livebirths). The commonest individual congenital anomaly was Down syndrome (1,33 per 1 000 livebirths), followed by neural tube defects (0,99 per 1 000 livebirths) and ventricular septal defects (0,69 per 1 000 livebirths). In 11% (2,25 per 1 000 livebirths) of neonatal deaths, infant loss was attributable to congenital anomalies.

Conclusions. The incidence of congenital anomalies in black South African neonates, born in an urban setting, is as high as in other First- and Third-World countries, and the incidence of some individual congenital anomalies is higher. This study indicates the need for further research and the establishment of prenatal, genetics and paediatric facilities to manage these problems.

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The World Health Organisation has indicated that it is necessary to evaluate the potential burden of congenital disorders in every country, whatever its stage of development, with a view to introducing preventive measures at the appropriate time.[1] As infectious diseases and malnutrition are brought under control in Third-World countries, congenital malformations will assume a greater relative importance as a cause of mortality and morbidity among infants and children, as has been the case in the First World.[2]

The incidence of congenital anomalies varies among different ethnic groups.[3,4] Extensive literature is available on the incidence of congenital anomalies in First-World countries[2-6] and studies have also been done in several Third-World countries.[7-10] However, in Africa south of the Sahara, limited information is available on the incidence of congenital anomalies, especially in the black population. Previous studies all had limitations in that they were either retrospective, performed on small sample numbers, or reported the frequency of a single abnormality or a few specific abnormalities.[10-14]

Only one prospective study on congenital anomalies in a significant number of liveborn black neonates has, to our knowledge, been reported previously. This was performed 20 years ago in Pretoria.[14]

We report the congenital anomalies profile of black liveborn neonates, delivered at a Pretoria hospital over a 3-year period. For the purposes of this study, the definition of a congenital anomaly was the same as that used by Christianson *et al.*[3] It encompasses conditions of prenatal origin, with structural defects, functional abnormalities, inborn errors of metabolism and chromosomal aberrations.

Patients and methods

This study was carried out in the neonatal unit and postnatal wards at Kalafong Hospital, an academic hospital that forms part of the academic medical complex of the University of Pretoria. By virtue of its location, this hospital serves mainly the black community to the west of Pretoria, but is also a referral hospital for the remainder of Pretoria and surrounding Northern Transvaal.

All liveborn babies born at the hospital between 1 May 1986 and 30 April 1989 were included in the study. A paediatrician examined every baby admitted to the neonatal unit, whatever the reason. Infants were routinely admitted to this unit for low birth weight, prematurity, any noted illness or abnormality, phototherapy and for assessment and routine observation following any form of delivery other than normal vaginal delivery. All normal liveborn babies, delivered by normal vaginal delivery, were admitted with their mothers to a postnatal ward, where within the first 24 hours of life they were examined by a medical officer attached to the paediatric department. In consequence, 6 853 (39,5%) infants were examined by a paediatrician and 10 498 (60,5%) by medical officers.

All diagnoses were recorded and assigned a code from the International Classification of Diseases (ICD-9).[15] In those cases where an infant had congenital abnormalities involving two or more systems, a diagnosis was made if possible, and the infant recorded only once under that code. If no diagnosis could be made, the infant was classified and coded as having a multiple congenital abnormality.

Departments of Paediatrics and Human Genetics and Developmental Biology, University of Pretoria

S. D. Delport, M.MED. (PAED.), M. PHARM. MED.

A. L. Christianson, M.R.C.P. (U.K.)

G. S. Gericke, M.D., F.C.P. (S.A.)

Genetic Services, Department of National Health and Population Development, Pretoria

H. J. S. van den Berg, R.N.

Institute for Biostatistics, Medical Research Council, Pretoria

L. Wolmarans, B.SC.

The number of liveborn infants delivered at Kalafong Hospital during this period was confirmed by examination of the maternity ward birth register. Infants with anencephaly were not referred to the neonatal unit or postnatal wards. The number of infants with this condition was obtained retrospectively from the birth register.

Postminimus polydactyly was prospectively excluded from the study, as this is a known common anomaly in black Africans and has no clinical significance for the patients.[10] Infants with congenital anomalies who died during the neonatal period were included in the study.

Because the study design was limited to the spectrum of clinical problems and their outcome in liveborn infants born at the hospital, congenital anomalies in stillborn infants were excluded; postmortems were not undertaken on the neonatal deaths from congenital anomalies because of limited pathological facilities.

Results

A total of 17 351 livebirths was delivered during the 3-year study period and 206 infants with congenital anomalies were recorded, resulting in an incidence of 11,87 per 1 000 livebirths.

The types of congenital anomalies are listed in descending order of frequency in Tables I-VI. They are broadly categorised into major systems, with incidence of specific defects per 1 000 livebirths.

Table I. Congenital anomalies — incidence and classification by system involved

Congenital anomaly and ICD-9 code	No.	%	Incidence per 1 000 livebirths
Central nervous system 7400-29, 3313-4, 3351, 33809	40	19,42	2,30
Musculoskeletal system 7540-69	37	18,0	2,13
Cardiovascular system 7450-79, 7593	31	15,1	1,79
Chromosomal 7580-82	29	14,1	1,67
Gastro-intestinal system 7490-519	19	9,22	1,09
Urogenital system 7520-539	16	7,80	0,92
Mendelian inheritance 2702, 2377, 7571-9, 75643, 75650, 3351	12	5,87	0,69
Multiple congenital abnormalities 7597	8	3,90	0,46
Haemangioma 2280	7	3,40	0,40
Integument 75738, 6948	2	0,97	0,12
Eye 7431-39	2	0,97	0,12
Other 77982-7440	2	0,97	0,12
Respiratory system 7480-89	1	0,48	0,06
Total	206	100	11,87

Table II. Congenital central nervous system anomalies

Congenital anomaly	ICD-9 code	No.	Incidence per 1 000 livebirths
Hydrocephalus	3313, 3314, 7423	11	0,63
Microcephalus	7421	8	0,46
Spina bifida without hydrocephalus	7419	6	0,35
Spina bifida with hydrocephalus	7410	5	0,29
Anencephaly*	7400	4	0,23
Encephalocele	7420	2	0,12
Porencephaly	74241	1	0,06
Cerebral cysts	34809	1	0,06
Spina bifida occulta	7561	1	0,06
Macrocephaly	7424	1	0,06

* No. of cases derived from review of maternity ward birth register.

Table III. Congenital musculoskeletal anomalies

Congenital anomaly	ICD-9 code	No.	Incidence per 1 000 livebirths
Congenital genu recurvatum	7544	9	0,52
Talipes equinovarus	75450	8	0,46
Anomalies of the diaphragm	7566	3	0,17
Skull, face, jaws	7540, 7560	3	0,17
Prune belly syndrome	75672	3	0,17
Arthrogryposis multiplex congenita	75580	2	0,12
Syndactyly	7551	2	0,12
Congenital dislocation of hip	7543	1	0,06
Spinal anomalies	7561	1	0,06
Absence of hand/fingers/ radius	7552	1	0,06
Talipes calcaneovalgus	7546	1	0,06
Absence of foot	7556	1	0,06
Anomalies of thoracic cage	7563	1	0,06
Unspecified cartilage anomaly	7509	1	0,06

Table IV. Congenital cardiovascular system anomalies

Congenital anomaly	ICD-9 code	No.	Incidence per 1 000 livebirths
Ventricular septal defect	7454	12	0,69
Patent ductus arteriosis*	7470	6	0,35
Atrial septal defect	7455	4	0,23
Pulmonary valve atresia	74600	2	0,12
Transposition of great arteries	7451	2	0,12
Hypoplastic left heart syndrome	7467	1	0,06
Cor biloculare	7457	1	0,06
Coarctation aorta	7471	1	0,06
Dextrocardia	7593	1	0,06
Complete heart block	4260	1	0,06

* 32 cases of patent ductus arteriosis in preterm infants not included.

Table V. Congenital gastro-intestinal system anomalies

Congenital anomaly	ICD-9 code	No.	Incidence per 1 000 livebirths
Inguinal hernia with/ without obstruction	5501 & 5509	6	0,35
Atresia and stenosis — small intestine	7511	4	0,23
Atresia and stenosis — large intestine, rectum, anus	7512	2	0,12
Cleft lip and palate	7492	2	0,12
Micrognathia	5240	2	0,12
Hiatus hernia	7506	1	0,06
Anomalies of mouth/pharynx	7502	1	0,06
Cleft lip	7491	1	0,06

Table VI. Congenital urogenital anomalies

Congenital anomaly	ICD-9 code	No.	Incidence per 1 000 livebirths
Hypospadias	7526	5	0,29
Undescended testes	7525	4	0,23
Unspecified anomalies of genital organs	753	2	0,12
Congenital posterior urethral valves	75360	1	0,06
Multicystic renal dysplasia	7531	1	0,06
Obstruction/atresia/stenosis of anterior urethra	75362	1	0,06
Unspecified anomalies of kidney	7533	1	0,06
Congenital renal failure	77980	1	0,06

Central nervous system congenital anomalies were the most frequent and accounted for 40 cases, giving an incidence of 2,30 per 1 000 livebirths (Table I). These were followed by congenital anomalies of the musculoskeletal system (2,13 per 1 000 livebirths), cardiovascular system (1,79 per 1 000 livebirths) and chromosomal abnormalities (1,67 per 1 000 livebirths). The gastro-intestinal system (1,09 per 1 000 livebirths) and urogenital system (0,92 per 1 000 livebirths), were the other systems with a high frequency of congenital anomalies.

The major congenital anomaly in the central nervous system (Table II) was hydrocephalus (0,63 per 1 000 live-births). Spina bifida, with or without hydrocephalus, had an incidence of 0,64 per 1 000 livebirths, and anencephaly an incidence of 0,23 per 1 000 livebirths. The incidence of neural tube defects, including encephalocele (0,12 per 1 000 livebirths), was thus 0,99 per 1 000 livebirths.

Congenital genu recurvatum and talipes equinovarus were the most commonly encountered musculoskeletal disorders (Table III). Postminimus polydactyly, a common anomaly in black Africans,[10] was not included in the study, as this anomaly has no clinical significance for the patients.

There was a total of 29 proven chromosomal disorders (1,67 per 1 000 livebirths). Down syndrome was the commonest of all diagnoses. The incidence of Down syndrome was 1,33 per 1 000 livebirths or 1 in every 751 babies delivered. The ages of the mothers of the 23 babies with Down syndrome ranged from 17 to 46 years (mean age 33 years) and 13 (52%) of the mothers were 35 years of age or older. Chromosomal confirmation was also obtained in 3 cases of trisomy 18 (0,17 per 1 000 livebirths), 2 cases of trisomy 13 (0,12 per 1 000 livebirths), and a single translocation trisomy 13 (0,06 per 1 000 livebirths).

Septation defects were the most frequent cardiovascular anomalies (Table IV) and undescended testes and hypospadias the commonest urogenital anomalies (Table VI). The incidence of cleft lip/palate was only 0,23 per 1 000 livebirths (Table V).

The integumentary conditions seen included congenital ichthyosis, epidermolysis bullosa, both disorders of Mendelian inheritance, an unclassifiable bullous dermatitis and a naevus.

Albinism was the most frequently diagnosed single gene disorder with an incidence of 0,23 per 1 000 livebirths or 1 affected neonate in every 4 350 deliveries. Other conditions due to Mendelian inheritance included 3 cases of neurofibromatosis (0,17 per 1 000 livebirths) and single cases of achondroplasia, osteogenesis imperfecta and spinal muscular atrophy (0,06 per 1 000 livebirths).

During the study a total of 354 neonates died during the neonatal period, giving a neonatal mortality rate of 20,4 per 1 000 livebirths. Of the neonates that died, 39 (11%) had a significant congenital anomaly. The neonatal mortality rate attributable to a birth defect was 2,25 per 1 000 livebirths. Twelve neonates (30% of those that died) had central nervous system defects, including 6 with neural tube defects and 4 with isolated hydrocephalus. Five neonates (12,8%) with congenital heart defects died, including 2 with pulmonary valve atresia. Other common causes of death were chromosomal abnormalities (4 neonates) and prune belly syndrome (3 neonates).

Discussion

The overall incidence of congenital anomalies (excluding postminimus polydactyly) in this study was 11,87 per 1 000 livebirths. This figure is almost certainly an underestimate, as the majority of babies in the study (60,4%) were examined once only by a medical officer, in the first 24 hours of life. However, this incidence is comparable to those recorded by Christianson *et al.*,[3] in a study done under more ideal First-World circumstances, and two recent similar studies from Third-World countries,[7,8] but is much lower than the incidence of congenital anomalies recently reported from Tunis.[9]

The incidence of congenital anomalies in the previous Pretoria study,[14] excluding postminimus polydactyly, was only 6,50 per 1 000 livebirths, which is significantly lower than in the present study. The reasons for the difference in total incidence could include the improved ability to detect abnormalities in the present study, because of increased awareness of congenital anomalies and the use of improved technology. It is also notable that in the first Pretoria study minor malformations such as cryptorchidism, mild hypospadias and skin malformations were not included.[14]

At the time this study was undertaken, facilities for prenatal screening and diagnosis in the community served by this hospital were non-existent. Therefore, pregnant women referred to the hospital for management were sent

for obstetric reasons, rather than for fetal abnormalities. Selection bias for congenital anomalies was therefore considered to be minimal.

In our study the percentage of deaths attributable to congenital anomalies (11%) was comparable to a recent similar study in Singapore (9,58%).[8] In the Pretoria study[14] the neonatal mortality rate was calculated at 14,9 per 1 000 livebirths and the neonatal mortality rate attributable to congenital anomalies was 1,01 per 1 000 livebirths. Thus 6,8% of neonatal deaths in that study were considered to be secondary to congenital anomalies. Given the improvement in pre- and perinatal management over the last 20 years, the neonatal mortality rate of the Pretoria study[14] is difficult to explain and should be regarded with caution. The suggested explanations of the difference in total congenital anomaly incidences may pertain to the neonatal mortality figures as well.

The system most commonly affected with congenital anomalies in this study was the central nervous system, which accounted for 40 (19,42%) cases. Isolated congenital hydrocephalus was the most common malformation noted in this group, with an incidence of 0,63 per 1 000 livebirths. It was also the commonest individual abnormality responsible for neonatal death. The high incidence of congenital hydrocephalus in black African neonates is a phenomenon which has been documented previously.[10,11] However, to our knowledge, no further information is available on the possible aetiological factors involved. Further research on this topic is thus required to clarify this issue. The incidence of congenital hydrocephalus in white neonates in Johannesburg is much lower (0,26 per 1 000 livebirths).[10]

Neural tube defects, anencephaly, spina bifida and encephaloceles had an incidence of 0,99 per 1 000 livebirths and, combined, were the commonest abnormality responsible for neonatal death. This incidence is similar to most previously published figures for black African urban populations in South Africa.[10] Ncayiyana,[13] however, described an incidence of neural tube defects of 6,13 per 1 000 livebirths in a rural black population of South Africa. The reason for such a high incidence of neural tube defects in this particular area remains unexplained. In comparison with other studies reported from the Third World, our incidence of neural tube defects was lower than previously recorded in Tunis (2,1 per 1 000 livebirths)[9] and Nairobi (2,65 per 1 000 livebirths),[12] but comparable to the incidence reported from Abu Dhabi (0,99 per 1 000 livebirths)[7] and higher than the incidence in Singapore (0,51 per 1 000 livebirths).[8] The figure is also comparable with most recorded incidences of neural tube defects in white neonates in South Africa.[10] Penrose[16] noted that anencephaly was rare in African people, which appears to be the case in this and most other studies reported from Africa.[10-12] It is possible that this finding is due to inadequate ascertainment of cases, as anencephalics are not routinely transferred to neonatal or postnatal wards.

The musculoskeletal system was the second commonest system affected by congenital anomalies. In previous studies from the Third World,[8-12] it is usually the most commonly affected system. Had postminimus polydactyly been included, the incidence of musculoskeletal abnormalities would have exceeded central nervous system abnormalities.

The cardiovascular system had the third-highest frequency of congenital anomalies, but cardiovascular malformations were the second commonest cause of neonatal death. The incidence of cardiovascular system anomalies was 1,79 per 1 000 livebirths, which was higher than in the studies reported previously from the Third World,[7-9,11,12] and significantly higher than the incidence of 0,20 per 1 000 livebirths reported in the previous Pretoria study.[14] However, the incidence is lower than that reported from First-World countries.[5] This incidence in our study indicates that the ascertainment and final diagnosis of cardiovascular abnormalities in this study were far more accurate than in the previous Pretoria study,[14] confirming the impression that there were many limitations to that study and that the research needed to be repeated.

Down syndrome was the commonest individual condition recorded in our study. All the patients diagnosed had non-disjunction trisomy 21. The incidence of Down syndrome was 1,33 per 1 000 livebirths or 1 in 752 babies delivered. Kromberg *et al.*,[17] in an ongoing study of Down syndrome in blacks, have reported an estimated interim incidence of 1,67 per 1 000 livebirths (1 in 600 babies). In the previous Pretoria study,[14] the incidence reported was 0,59 per 1 000 livebirths, but it was noted that Down syndrome was much less readily detectable on superficial examination in some ethnic groups, and that, unless careful clinical examination was undertaken in the newborn and supplemented by chromosome analysis, studies of birth frequency had to be interpreted with caution.[14] This latter figure[14] is therefore considered to be a gross underestimation of the true Down syndrome incidence, and it is possible that the incidence in our study, and the estimate of Kromberg *et al.*,[17] are lower than the true incidence of Down syndrome in black African populations, because of incomplete ascertainment.

Kromberg *et al.*[17] noted that 55% of mothers of Down syndrome babies in their study were over 35 years of age, but that none of these women had been offered prenatal diagnosis. Similarly, in our study 52% of the mothers of Down syndrome infants were over the age of 35 years. These figures indicate the necessity in South Africa for counselling clinics and the attendant support of family planning, genetics and obstetric services for black women of advanced maternal age.

Cleft lip and/or palate had a very low incidence (0,23 per 1 000 livebirths), a phenomenon which has previously been recorded.[10]

Albinism and neurofibromatosis were the two commonest single-gene disorders diagnosed in the study. The incidence of albinism was 0,23 per 1 000 livebirths, which is similar to the prevalence previously reported by Kromberg *et al.*[18] No previous incidence of neurofibromatosis in black Africans had, to our knowledge, been calculated, and the incidence of 0,17 per 1 000 livebirths thus possibly represents a minimal incidence in African people.

Conclusion

This study was undertaken as part of a larger study to assess the clinical spectrum and outcome of problems in neonates born at Kalafong Hospital. In consequence, the incidence of congenital anomalies could be ascertained.

The overall incidence of congenital anomalies in black African neonates was shown to be at least as high as, and in some instances higher than, in other countries and other ethnic groups. Our study highlights the paucity of information on congenital anomalies in black African populations and the need for further elucidation of their incidence and causative mechanisms. The information derived from such studies could facilitate planning for the provision of future family planning, prenatal, genetics and paediatric services, and the initiation of specific programmes to reduce the incidences of selected common anomalies, such as Down syndrome, neural tube defects and congenital hydrocephalus.

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REFERENCES

1. Department of National Health and Population Development, Genetic Services Division. *National Birth Defects Surveillance System Annual Report*. Pretoria: Government Printer, 1991.
2. Kennedy WP. Epidemiological aspects of the problem of congenital malformations. *Birth Defects* 1967; **3**(2): 1-18.
3. Christianson RE, Van den Bergh BJ, Milkovich L, Oechsli FW. Incidence of congenital anomalies among white and black live births with long-term follow up. *Am J Public Health* 1981; **71:** 1333-1341.
4. Terry PB, Bissenden JG, Condie RG, Matthew PM. Ethnic differences in congenital malformations. *Arch Dis Child* 1986; **60:** 866-879.
5. Lumley J, Palma S, Fisher M, Robertson H. The tip of the iceberg: a validation study of the Victoria congenital malformation register. *Aust Paediatr J* 1988; **24:** 244-246.
6. Van Regemorter N, Dodion J, Druart C, *et al*. Congenital malformations in 10 000 consecutive births in a university hospital: need for genetic counselling and prenatal diagnosis. *J Pediatr* 1984; **104**(3): 386-390.
7. Al-Jawad ST, Shubber AI, Khafaji NA, Kholeif SA, Lane SM, Moore DK. A survey of serious congenital morphological abnormalities in Abu Dhabi. *Ann Trop Paediatr* 1988; **8:** 76-79.
8. Thein MM, Koh D, Tan KL, *et al*. Descriptive profile of birth defects among livebirths in Singapore. *Teratology* 1992; **46:** 277-284.
9. Khrouf N, Spang R, Podgorna T, Miled SB, Moussaoui M, Chabani M. Malformations in 10 000 consecutive births in Tunis. *Acta Paediatr Scand* 1986; **74:** 534-539.
10. Kromberg JGR, Jenkins T. Common birth defects in South African blacks. *S Afr Med J* 1982; **62:** 599-602.
11. Simpkiss M, Lowe A. Congenital abnormalities in the African newborn. *Arch Dis Child* 1961; **36:** 404-406.
12. Khan AA. Congenital malformations in African neonates in Nairobi. *J Trop Med Hyg* 1965; **68:** 272-274.
13. Ncayiyana DJ. Neural tube defects among rural blacks in a Transkei district. *S Afr Med J* 1986; **69:** 618-620.
14. Stevenson AC, Johnson HA, Stewart MIP, Golding DR. Congenital malformations: a report of a series of consecutive births in 24 centres. *Bull World Health Organ* 1966; **34:** suppl, 1-127.
15. *International Classification of Diseases*. Vol 1. 9th ed. Geneva: World Health Organisation, 1977.
16. Penrose LS. Genetics of anencephaly. *J Ment Defic Res* 1957; **1:** 1.
17. Kromberg JGR, Christianson AL, Duthie-Nurse G, Zwane E, Jenkins T. Down syndrome in the black population (Letter). *S Afr Med J* 1992; **81:** 337.
18. Kromberg JGR, Jenkins T. Prevalence of albinism in the South African Negro. *S Afr Med J* 1982; **61:** 385-386.

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