REDUCTION DEFORMITIES, TWINNING AND MORTALITY IN BRAZILIAN WHITES AND NEGROES

N. FREIRE-MAIA, J.B.C. AZEVEDO

Department of Genetics, Federal University of Paraná, Curitiba, Paraná, Brazil; and School of Philosophy, Marilia, SP, Brazil.

A series of house-to-house surveys in Central Brazil revealed 32 cases of reduction deformities of the limbs (brachydactyly, synbrachydactyly, ectrodactyly, amely, adactyly, achiry, apody, etc.) among 58,761 births (0.54 per thousand, being 0.66 per thousand among Whites and 0.43 per thousand among Mulattoes and Negroes). No sibship was found with two or more equally affected singleton members. One case of concordance among twins is referred. There were no parity, inbreeding, race and sex effects. Both hands and both feet were also equally affected. Precocious mortality and twinning were significantly higher among affected individuals. The presence of cleft lip and palate in one of our index patients may also reflect an etiological relationship. Among the sibs, the total incidence of congenital malformations was similar to that generally accepted for general populations.

INTRODUCTION

Reduction deformities of the limbs are a rather heterogeneous nosologic group both as regards clinical and etiological aspects (Freire-Maia 1969). In the large majority of them, etiology is totally unknown and the average recurrence risk is practically nil. The present investigation (made on the basis of a house-to-house survey) had the advantage of avoiding ascertainment biases which would increase recurrence, unlike our first investigations (Freire-Maia and Freire-Maia 1964 and 1967, Fonseca and Freire-Maia 1970). Some of its main conclusions have already been briefly mentioned before (Freire-Maia and Azevedo 1968, Freire-Maia 1969).

MATERIAL AND METHODS

The surveys from which the present data emerged have been made with the aim of obtaining data for inbreeding studies and fully described before (Freire-Maia et al. 1963, Freire-Maia 1963, Freire-Maia and Azevedo 1971, Krieger et al. 1971). They have been made in the south of the central Brazilian State of Minas Gerais, among socioeconomic levels from medium to very low, with predominance of the low levels where cooperation proved to be higher. In some of the surveys, the families were ascribed numbers which would correspond approximately to socioeconomic levels, from 5 (the highest) to 1 (the lowest). The mean was equal to 1.68 ± 0.01, thus showing a rather low average social status (Pederneiras et al. 1974).

The affected individuals not present at home at the time of the interviews were described (generally by their own mother) as accurately as possible. Long descriptions were sometimes obtained in order to get as detailed information as possible. When the affected individuals were present, they were examined by the interviewers. When necessary, drawings were made of the affected parts. In some cases, photographs were taken, and in one case, a radiograph.

CODEN: AGMGAK 26 133 (1977) — ISSN: 0001-5660
Acta Genet. Med. Gemellol. (Roma), 26: 133-140

The first survey has been made directly by the authors; the others, by duly instructed persons under the direct supervision of the authors.

We define " reduction deformities " in this paper as any congenital malformation due either to aplasias or to minor or major hypoplasias of one or more bones of the limbs. Our list of malformations in the next section shows the large range investigated, from mild brachydactylies and synbrachydactylies to some of the most severe reduction deformities, such as phocomelias, amelias, achiries, etc.

THE DATA

A total of 58,761 births (including stillbirths) have been ascertained with a total of 32 cases of reduction deformities (0.54 per thousand). The incidence among White (19/28,809 = 0.66 per thousand) is statistically not significantly different ($\chi_1^2 = 1.37$; $p > 0.20$) from that among non-Whites (i.e., Mulattoes and Negroes) (13/29,952 = 0.43 per thousand). All the relatives of our index patients are normal, except when specified below.

No sibship was found with two or more similarly affected singleton members. A total of 95 births occurred after the birth of our index patients.

A short description of each malformation, as classified by race, sex, and F (the inbreeding coefficient of the affected individual) will be presented below. When F is not mentioned, this will mean that it is equal to 0.

Whites

Males

1. *Brachydactyly* (with absence of nails; right hand and both feet; died at 40 days).
2. *Poland paradrome* (= anomalad) (right side affected). (For details on this patient, see Freire-Maia et al. 1973).
3. *Brachydactyly* (hypophalangy of the fifth left toe; died at one day).
4. *Brachydactyly* (lower unilateral).
5. *Ectrodactyly* (absence of two toes on both feet; $F=5/64$).
6. *Ectrodactyly* (absence of the first and fifth fingers of the left hand; died on the first day).
7. *Apody* (right; associated with imperforate anus; died at 18 days).
8. *Amputation* (quadruple above knee and above elbow; died at one day; $F=1/32$).
9. *Phocomelia* (complete lower bilateral; stillborn).
10. *Ectrodactyly and brachymetacarpy* (right hand; absence of the first, second, fourth and fifth fingers; associated with short corresponding metacarpals).
11. *Achiry* (left; $F=1/16$).

Females

1. *Ectrodactyly* (quadruple; ring constrictions in fingers and toes as well as in the right leg).
2. *Brachydactyly* (hypophalangy of second and third toes of the right foot).
3. *Ectrodactyly and syndactyly* (absence of third finger and syndactyly of fourth and fifth fingers of the left hand).
4. *Achiry* (left; $F=1/16$).
5. *Amelia* (upper bilateral; died at three days).
6. *Adactyly* (left hand).
7. *Brachydactyly* (bilateral hypophalangy of the third fingers and toes associated with harelip and cleft palate; died at three months; a cotwin is a normal female; $F=1/16$).
8. *Ectrodactyly and brachydactyly* (absence of two right fingers; fifth finger shorter).

MULATTOES AND NEGROES

Males

1. *Poland paradrome* (left side affected). (For details on this patient, see Freire-Maia et al. 1973).
2. *Brachydactyly* (hypoplastic second toe of both feet).
3. *Brachydactyly* (hypoplastic second toe of both feet).
4. *Ectrodactyly* (left hand; absence of first, fourth and fifth fingers; a brother with bilateral fibular polydactyly).
5. *Ectrodactyly* (absence of two toes on the left foot; a cotwin is a normal female).

Females

1. *Brachydactyly* (hypophalangy in all fingers of the left hand).
2. *Synbrachydactyly* (bilateral webbed toes; hypoplastic second toe of both feet; a cotwin is the following (no. 3).
3. *Synbrachydactyly* (bilateral webbed toes; hypoplastic second toe of the right foot; a cotwin is that described under no. 2).
4. *Phocomelia* (incomplete upper bilateral; stillborn; a cotwin is a normal male).
5. *Ectrodactyly and syndactyly* (right splithand with three fingers and syndactyly of the fourth and fifth; syndactyly of second to fifth fingers of left hand; only three toes on the right foot).
6. *Ectrodactyly* (absence of right thumb; died at one day).
7. *Ectrodactyly* (absence of one toe on the left foot; a sister with harelip and cleft palate).
8. *Achiry* (left).

Among the above 31 sibships with 32 patients, 5 were the offspring of consanguineous marriages ($16.13 \pm 6.61\%$). In a sample from the general population where the patients have been ascertained, the frequency of consanguineous marriages was found to be $1152/10{,}260 = 11.23 \pm 0.31\%$. These two frequencies are not statistically different ($\chi_1^2 = 0.75$; $p > 0.30$). Interestingly enough, all the 5 inbred sibships are White ($5/19 = 26.31 \pm 10.10\%$). In our subsample from the White fraction of the general population, the frequency of consanguineous marriages ($743/4959 = 14.98 \pm 0.51\%$) is not statistically different from 26% ($\chi_1^2 = 1.92$; $p > 0.10$). The frequency of inbred sibships among Whites (5/19) is also not statistically different from 0/12 among non-Whites ($\chi_1^2 = 3.77$; $p > 0.05$). The inbred patients had lower bilateral ectrodactyly (1 case), quadruple amputation (1 case), left achiry (2 cases) and bilateral hypophalangy of the third fingers and toes associated with cleft lip and palate (1 case). Among the 32 affected indviduals, 1 was stillborn (complete lower bilateral phocomelia), 5 died in the first week of life (hypophalangy of the fifth left toe; absence of the first and fifth fingers of the left hand; quadruple above knee and above elbow amputation; upper bilateral amelia; absence of the right thumb); 1 died in the third week of life (right apody); 1 died in the second month of life (brachydactyly of the right hand and both feet associated with absence of nails), and 1 died at three months (bilateral hypophalangy of the third fingers and toes associated with cleft lip and palate). These 9 cases represent $28.13 \pm 7.93\%$ of the 32 affected individuals. (In the subsequent comparisons, Yates' correction for continuity will be always applied when justifiable.) This frequency (28%) is statistically different from the corresponding mortality among their sibs ($19/189 = 10.05 \pm 2.19\%$; $\chi_1^2 = 6.53$; $p < 0.05$). Of the 9 cases of mortality (from stillbirth to the third month of life), 8 are among Whites ($8/19 = 42.11 \pm 11.33\%$), whereas the mortality among their sibs is $9/109 = 8.26 \pm$

2.64% ($\chi_1^2 = 13.29$; $p<0.01$) and only 1 (with absence of the right thumb) is non-White ($1/13 = 7.69 \pm 7.39\%$), whereas the mortality among their sibs is $10/80 = 12.50 \pm 3.70\%$ ($\chi_1^2 = 0.25$; $p>0.50$). The difference between the mortality of the affected Whites (42%) and that of the non-Whites (8%) as well as the difference between those of their sibs (8% and 13%) are not significant (χ_1^2 equals 3.43 and 0.92, respectively; $p>0.05$ and >0.30, respectively).

If we classify the 32 anomalies into different types on the basis of generally accepted criteria, we have the incidences as shown in the Table. There are no suggestions that these incidences may vary between the two ethnic groups (all the χ^2 values correspond to values of p higher than 0.05). Fifteen of the malformations affected the upper limbs, 12 the lower limbs and 5

Table. *Incidence of different types of reduction deformities*

	Whites		Non-Whites		Total
Deformity[a]	Per 28,809	Per 10,000	Per 29,952	Per 10,000	Per 10,000
Amelia	1	0.35	0	0	0.17
Achiry	2	0.69	1	0.33	0.51
Phocomelia	1	0.35	1	0.33	0.34
Brachydactyly[b]	5	1.74	3	1.00	1.36
Ectrodactyly only	3	1.04	4	1.34	1.19
Ectrodactyly[c]	3	1.04	1	0.33	0.68
Amputation	1	0.35	0	0	0.17
Adactyly	1	0.35	0	0	0.17
Apody	1	0.35	0	0	0.17
Synbrachydactyly	0	0	2	0.67	0.34
Poland paradrome	1	0.35	1	0.33	0.34
Upper limbs	9	3.12	6	2.00	2.55
Lower limbs	6	2.08	6	2.00	2.04
Upper and lower	4	1.39	1	0.33	0.85
Total	19	6.60	13	4.34	5.45
Hand defects[d]	11	3.82	6	2.00	2.89
Foot defects[d]	8	2.78	7	2.34	2.55
Hand def. only	7	2.43	4	1.34	1.87
Foot def. only	4	1.39	6	2.00	1.70

[a] As described in the list presented in *The data;* [b] One associated with cleft lip and palate; [c] Associated with other defects; [d] With and without other defects.

both of them ($\chi_2^2 = 4.95$; $p>0.05$). The incidence of patients with both upper and lower limbs affected (0.85/10,000) is about 1600 times higher than would be expected by mere chance on the basis of the product of the incidence of the reductions of the upper limbs alone and that of the lower limbs alone.

Males and females are equally affected (16 cases each). Both sides are also equally affected: 5 right hands and 9 left hands ($\chi_1^2 = 1.14$; $p>0.20$) and 3 cases in each foot.

Among the 31 pregnancies that resulted in at least one individual affected with reduction deformities, 4 led to twins ($12.90 \pm 6.02\%$), a high frequency as compared with that from

the same couples but resulting in " normal " children, i.e., not affected with reduction deformities ($2/187 = 1.07 \pm 0.75\%$; $\chi_1^2 = 9.84$; $p<0.01$). The twinning rate as regards affected Whites is $1/19 = 5.26 \pm 5.10\%$, while that as regards affected non-Whites is $3/12 = 25.00 \pm 12.48\%$ ($\chi_1^2 = 1.10$; $p>0.20$). The twinning rate among their " normal " sibs equals 0/109 for Whites and $2/78 = 2.56 \pm 1.78\%$ for non-Whites ($\chi_1^2 = 0.92$; $p>0.30$). The difference between affected and " normal " Whites is also nonsignificant ($\chi_1^2 = 0.99$; $p>0.30$), but that between affected and " normal " non-Whites is significant ($\chi_1^2 = 6.16$; $p<0.05$).

In Brazilian general populations, the twinning rates have been found to range between 0.97% and 2.42%, the majority lying between 1.19% and 1.75%, without consistently showing Negroid populations with higher rates than Caucasians (Arena 1974, Pedreira et al. 1959, Freire-Maia et al. 1961, Souza et al. 1966, Saldanha et al. 1963, Stevenson et al. 1966, Araújo and Salzano 1975, Azevedo and Freire-Maia 1970). The data by Freire-Maia et al. (1961) and Azevedo and Freire-Maia (1970), obtained in the same surveys on which our present data on reduction deformities are based, led to 1.19%, practically identical to that we now found in our " control " group (1.07%).

There is no birth-order effect in our cases of reduction deformities. According to Haldane and Smith's method (1948), the sum of the birth order of the index patients equals 666 and its expected value on the assumption of no effect is 756. The difference between both values (90) is lower than twice the standard deviation, which is 77.38. According to Slater's method (1962), the average birth order is 0.58 ± 0.08, not statistically different from 0.50 ($t = 0.80$; $p>0.20$).

DISCUSSION

Since, as far as we know, no other research has employed exactly the same definition of reduction deformities and/or collected the data according to the same methodology, we think any comparison between our data and the others available in the literature would be worthless. Any possible differences and similarities could be due to unrelated concomitant variables. Our data only permit internal comparisons and characterize a specific population as investigated with the specific method which was used.

In spite of that, we would like to mention that other Brazilian populations have been investigated on the basis of clinical examinations of newborn children. The incidences of reduction deformities varied as follows: 0, 0.49, and 1.65 per thousand (Araújo and Salzano 1975, Stevenson et al. 1966, Arena 1974). In spite of the fact that the methodology employed by these authors differed very much from ours and is supposed to be better than it, our estimate (0.54 per thousand) occupies a central position rather than the lowest one, occupied by that of Araújo and Salzano, who investigated a total of 4900 Whites and 1067 non-Whites, all of them living at the time of the examination. Arena (1974), whose series of 3028 births also included 64 stillbirths, found 17 cases of " more proximal implantation of toes ", some of which could be due to metatarsal reduction (Arena 1976, pers. communic.).

All the reduction defects were sporadic. This confirms previous investigations which reported no recurrence at all among sibs for the large majority of reduction deformities (Birch-Jensen 1949, Freire-Maia and Freire-Maia 1964 and 1967, Fonseca and Freire-Maia 1970, Rogala et al. 1974). This is an optimistic aspect (for counseling purposes) which is coupled with the fact that etiology is generally unknown. For parents seeking counseling, therefore, the know-

ledge of the etiology of the anomaly which is present in one of their children is not always the best source of " tranquility ", whereas for the counselor it is. As in the present situation, complete ignorance of etiology may be associated with excellent genetic prognosis.

The high incidence (4/31) of twinning among the pregnancies which resulted in the index patients — a fact already mentioned by Birch-Jensen (1949) — points to the hypothesis that " exogenous " factors may have some etiological importance. Our data do not seem to suggest, however, that poor living conditions and factors associated with parity and race may be preponderant.

Our data also show no inbreeding effect, which, together with nonrecurrence, rules out the action of rare recessive genes. The fact that both sexes are equally affected shows that both are equally sensitive to the (unknown) etiological agents.

The incidence of *other* malformations among the sibs of our index patients is 2/189 ($1.06 \pm 0.74\%$), a value of the same order of magnitude as those of general populations. The face value is actually lower than those available for Brazilian (mixed) populations, where the reported incidences of congenital malformations, as ascertained on the basis of both prospective and retrospective approaches, vary from 2.00% to 16.09% (the majority being between 2% and 4%), the incidences of the major ones ranging from 1.27% to 2.46% (Saldanha et al. 1963, Araújo 1963, Saldanha 1964, Stevenson et al. 1966, Arena 1974, Araújo and Salzano 1975). This is a clear suggestion that reduction deformities have no etiological relationship with " other " malformations, i.e., the sibs of patients with reduction deformities do not seem to be more liable to develop other malformations than the average individual in general populations. Our present data are in full accordance with the conclusions of other investigations we have made into the same subject (Freire-Maia and Freire-Maia 1964 and 1967, Fonseca and Freire-Maia 1970).

In our sample, reduction deformities have been found associated with other malformations in a few instances: with imperforate anus (1 case), ring constrictions (1 case), syndactyly (4 cases), cleft lip and palate (1 case), and deficiency of thoracic muscles (Poland paradrome, 2 cases). It is possible that some of these associations are purely coincidental, but some of them (e.g., ring constrictions and syndactyly) are probably the result of the same etiological agent. The presence of one case of cleft lip and palate among our 32 index patients may also have an etiological significance, as shown by Birch-Jensen (1949). Poland paradrome (anomalad) is a well-known entity; our two cases have been studied in another paper (Freire-Maia et al. 1973). We estimated its incidence, on the basis of an indirect approach, as lying between 10^{-5} and 10^{-4}. In our data, its frequency is 1/30,000.

It is worth calling attention to the fact that among the 4 cases of twin pairs referred to in our list, 1 led to an almost complete concordance (synbrachydactyly; non-White females). Since only 2 of the 4 twin pairs have the same sex (♀♀) and, therefore, can be MZ, concordance among possible MZ twins is 1/2. This concordance bears a suggestion that the synbrachydactyly referred to in our list may be genetic.

It is doubtful whether the inbred sibships (or at least some of them) show malformations due to autosomal recessive genes. Since no recurrence occurred in any of them and the inbreeding rate is not significantly higher in the sample of index patients as compared with the general population, we prefer to assume that inbreeding is here a merely coincidental finding. Since Whites living in the Brazilian region surveyed have a Portuguese origin, one would be tempted to identify uncritically the quadruple amputation verified in an inbred ($F = 1/32$) White male patient as acheiropodia. This would not be supported by what is known as

regards the lower amputation level (always below the knee) in acheiropodia (Freire-Maia A. 1974 and 1975).

As mentioned, all 5 inbred index patients and 8 out of 9 of the cases of precocious mortality were found among Whites. There is, however, no apparent association between inbreeding and precocious mortality. The malformations verified among Whites are, on average, more severe than those of the non-Whites and this seems to be the cause of the higher precocious mortality of affected Whites. However, only 2/8 of the dead were inbred.

Acknowledgments

The authors wish to acknowledge the support given to this investigation by the Brazilian agencies CNPq and CAPES, by the Rockefeller Foundation and by WHO.

REFERENCES

Araújo J. 1963. Malformações congênitas. Pediatr. Prat., 34: 131-138. Cited by Araújo and Salzano 1975.

Araújo A.M. de, Salzano F.M. 1975. Congenital malformations, twinning and associated variables in a Brazilian population. Acta Genet. Med. Gemellol., 24: 31-39.

Arena J.F.P. 1974. Estudo clínico-epidemiológico prospectivo das anomalias congênitas na população de Campinas, SP. M.D. thesis, University of Campinas, Brazil.

Azevedo J.B.C., Freire-Maia N. 1970. Cited by Salzano and Freire-Maia 1970.

Birch-Jensen A. 1949. Congenital deformities of the upper extremities. In: Opera ex Domo Biologiae Hereditariae Humanae Universitatis Hafniensis, Vol. 19. Copenhagen: Ejnar Munksgaard.

Fonseca L.G., Freire-Maia N. 1970. Congenital malformations of limbs. Lancet, 1: 90-91.

Freire-Maia A. 1974. Genética da aquiropodia (" the handless and footless families of Brazil "). II. Aquiria e casos " similares " à aquiropodia. Rev. Paul. Med., 84: 107-110.

Freire-Maia A. 1975. Genetics of acheiropodia (" the handless and footless families of Brazil "). VIII. Penetrance and expressivity. Clin. Genet., 7: 98-102.

Freire-Maia N., Freire-Maia A., Quelce-Salgado A. 1961. A incidência de gêmeos, segundo o grupo étnico e a consangüinidade, em zona rural de Minas Gerais. Atas Prim. Sim. Sul-Americ. Genét. (Brazil), 266-267.

Freire-Maia N. 1963. The load of lethal mutations in White and Negro Brazilian populations. II. Second survey. Hum. Hered., 13: 199-225.

Freire-Maia N., Freire-Maia A., Quelce-Salgado A. 1963. The load of lethal mutations in White and Negro Brazilian populations. I. First survey. Hum. Hered., 13: 185:198.

Freire-Maia N., Freire-Maia A. 1964. Multiple congenital malformations. Lancet, 1: 113-114.

Freire-Maia N., Freire-Maia A. 1967. Recurrence risks of bone aplasias and hypoplasias of the extremities. Acta Genet., 17: 418-421.

Freire-Maia N., Azevedo J.B.C. 1968. Skeletal limb deficiencies. Lancet, 2: 1296.

Freire-Maia N. 1969. Congenital skeletal limb deficiencies. A general view. Birth Defect., Orig. Artic. Ser., 5, pt. 3: 7-13.

Freire-Maia N., Azevedo J.B.C. 1971. The inbreeding load in Brazilian Whites and Negroes as estimated with sib and cousin controls. Am. J. Hum. Genet., 23: 1-7.

Freire-Maia N., Chautard E.A., Opitz J.M., Freire-Maia A., Quelce-Salgado A. 1973. The Poland syndrome - Clinical and genealogical data, dermatoglyphic analysis, and incidence. Hum. Hered., 23: 97-104.

Haldane J.B.S., Smith C.A.B. 1948. A simple exact test for birth-order effect. Ann. Eugen., 14: 117-124.

Krieger H., Freire-Maia N., Azevedo J.B.C. 1971. The inbreeding load in Brazilian Whites and Negroes: Further data and a reanalysis. Am. J. Hum. Genet., 23: 8-16.

Pederneiras M.P., Karam Jr. E., Freire-Maia N. 1974. Consanguineous marriages and umbilical tetanus in Brazilian populations. Hum. Hered., 24: 75-81.

Pedreira C.M., Peixoto L.I.S., Rocha L.M.G.I. 1959. Estudo da gemelaridade na população de Salvador, Bahia. An. Prim. Reun. Brasil. Genét. Hum (Brazil), 137-140.

Rogala E.J., Wynne-Davies R., Littlejohn A., Gormley J. 1974. Congenital limb anomalies: Frequency and aetiological factors. Data from the Edinburgh Register of the newborn (1964-68). J. Med. Genet., 11: 221-233.

Saldanha P.H., Cavalcanti M.A., Lemos M.L. 1963. Incidência de defeitos congênitos na população de São Paulo. Rev. Paul. Med. 63: 211-229.

Saldanha P.H. 1964. Frequency of congenital mal-

formations in mixed populations of Southern Brazil. Proc. 2nd Int. Conf. Cong. Malf., Int. Med. Congress, New York [pp. 323-333].
Salzano F.M., Freire-Maia N. 1970. Problems in Human Biology. A Study of Brazilian Populations. Detroit: Wayne State University Press.
Slater E. 1962. Birth order and maternal age of homosexuals. Lancet, 1: 69-71.
Souza J.P., Andrade A.M.T.L., Ronzani D.M., Guimarães C.S., David J.A. 1966. Estudo estatistico do parto gemelar. Rev. Ginecol. Obstet., 119: 68-87. Cited by Araújo and Salzano 1975.
Stevenson A.C., Johnston H.A., Golding D.R., Stewart M.I.P. 1966. Comparative study of congenital malformations. Medical Research Council, Great Britain.

RIASSUNTO

Deformità Riduttive, Gemellarità e Mortalità nelle Popolazioni Bianca e Negra del Brasile

Una inchiesta domiciliare nel Brasile Centrale ha rivelato 32 casi di deformità riduttive degli arti (brachidattilia, sinbrachidattilia, ectrodattilia, amielia, adactilia, achiria, apodia, ecc.) su di un totale di 58.761 nati (0,54 per mille, 0,66 nei bianchi e 0,43 in negri e mulatti). Non si sono trovate fratrie con due o più membri mononati affetti e viene riferito un solo caso di concordanza in gemelli. Non si sono trovati effetti di parità, endogamia, razza o sesso. Le due mani ed i due piedi sono risultati ugualmente colpiti. Nelle famiglie degli individui colpiti mortalità precoce e gemellarità sono risultate significativamente più elevate. La presenza di cheilognatopalatoschisi in uno dei pazienti indice può anche riflettere una relazione eziologica. La frequenza complessiva di malformazioni congenite fra fratelli è risultata simile a quella accettata per la popolazione generale.

RÉSUMÉ

Deformités Réductionnelles, Gémellarité et Mortalité dans les Populations Blanche et Noire du Brésil

Une enquête domiciliaire dans le Brésil Central a révélé 32 cas de déformités réductionnelles des membres (brachydactylie, synbrachydactylie, ectrodactylie, amyélie, adactylie, achyrie, apodie, etc.) sur un total de 58.761 nés (donc 0,54 pour mil, avec 0,66 chez les blancs et 0,43 chez les noirs et les mulâtres). Des concordances entre frères n'ont pas été trouvées, à l'exception d'un cas de concordance chez deux jumeaux. Aucun effet de parité, endogamie, race, sexe ou latéralité n'a été trouvé. Mortalité précoce et gémelliparité étaient significativement plus élevées chez les familles des sujets atteints. La présence de cheilognathopalatoschise chez un des patients index pourrait refléter une relation étiologique. La fréquence de malformations congénitales chez les frères et sœurs n'était pas différente par rapport à la population générale.

ZUSAMMENFASSUNG

Reduzierende Missbildungen, Zwillingshäufigkeit und Sterblichkeit bei den Weissen und Schwarzen Populationen Brasiliens

Eine in Form von Hausbesuchen durchgeführte Forschung in Zentralbrasilien ergab 32 Fälle von reduzierender Missbildung der Gliedmassen (Brachydaktylie, Synbrachydaktylie, Ektrodaktylie, Amyelie, Adaktylie, Achirie, Apodie usw.). Dies entsprach 0,54‰ aller untersuchten Kinder (58.761): 0,66‰ der weissen und 0,43‰ der Neger- oder Mulattenkinder. Es fanden sich keine Familien, in der zwei oder mehr Kinder von den gleichen Missbildungen betroffen waren und es wird auch nur ein Fall von Zwillingskonkordanz erwähnt. Parität, Endogamie, Rasse oder Geschlecht scheinen das Phänomen nicht zu beeinflussen. Beide Hände und beide Füssen sind in gleichem Masse betroffen. In den Familien der Betroffenen fallen frühe Sterblichkeit und Neigung zu Zwillingsgeburten auf. Der Befund eines Wolfsrachens (Cheilognathopalatoschisis) bei einem der Indexpatienten kann auf einen etiologischen Zusammenhang hinweisen. Die Gesamthäufigkeit der angeborenen Missbildungen bei Geschwistern entsprach ungefähr der für die Gesamtbevölkerung angenommenen Schätzung.

Prof. Newton Freire-Maia, Departamento de Genética (UFPr), Caixa Postal AA, 80000 Curitiba, Paraná, Brasil.