Abstract

**Objective:** to identify and compare cases of isolated polydactyly with healthy newborns regarding familial and maternal characteristics.

**Methods:** we conducted this hospital-based case-control study from 1990 to 1998. We collected data in interviews with mothers of cases and controls during the post-delivery period. Cases were defined as newborns presenting an extra or a bifid digit in hands and/or feet. Controls were the four healthy newborns that were born after the case, at the same hospital. Information was gathered on type of polydactyly, gender and birthweight, twin pregnancy, parental consanguinity, ethnicity.

**Results:** African ancestry and positive family history of congenital malformation, especially polydactyly, were significantly associated with the occurrence of this type of congenital anomaly (crude OR of 3.3; 10.0; and 55.0, respectively).

**Conclusion:** isolated polydactyly was one of the most frequent malformations found in the studied population. The findings associated with black African ancestry confirm the data presented in the literature. Studies conducted on this same population, with a larger population of patients with this type of malformation, could investigate this association better and justify the results presented here.


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**Introduction**

In general, there are several cases of congenital anomalies among humans; and each of these cases has its own characteristics in terms of incidence, genetic influence, severity, and associated morbidity. Polydactyly, a very frequent malformation, is clinically characterized by the appearance of an extra digit in the hands and/or feet, when there is excessive radial division, or large or bifid digit when division is incomplete.\(^1\) These anomalies are classified as postaxial or preaxial polydactyly according to their respective manifestation on the ulnar/fibular or radial/tibial face of the hands or feet; and may concomitantly occur on both limbs and in both sides of the body.\(^2\)

The hereditary aspect (cause) of this malformation behaves differently according to geographical and consequently racial variations; as a result, the occurrence of extra digits manifests itself in varied ways.\(^1\) In the city of Pelotas (State of Rio Grande do Sul- Brazil), isolated polydactyly is the second most frequent congenital anomaly,\(^3\) with a prevalence of 15.6 cases per 10,000 births. The present study aims at tracing out a profile of the behavior presented by this isolated malformation (which is not associated with syndromes) in the population of the city of Pelotas.
Materials and methods

The study was carried out in the 5 existing maternity hospitals in Pelotas, a city with approximately 300,000 inhabitants, located in southern Brazil, through a period of 9 years (1990 through 1998). The ECLAMC (Latin American Collaborative Study of Congenital Malformations) questionnaire was used for data collection. This questionnaire was applied, right after delivery, to all mothers who gave birth to infants with polydactyly unrelated to other malformations. Four healthy individuals born in the same hospitals after the birth of any infants with some kind of malformation were used as control. Seventy-five percent of the control individuals were paired up with the cases of other malformations according to gender. The same questionnaire was applied to the mothers of control individuals.

The following variables were analyzed: types of polydactyly, gender, weight, mother’s age, twin pregnancy, parental consanguinity, number of pregnancies, mother’s level of education, ethnicity, and family history of malformations.

Polydactyly types were categorized as postaxial and preaxial, and subdivided according to their location (hands, feet, or both).

Gender and weight were obtained from the maternity ward records. Mothers’ ages are expressed in years. Twin pregnancy refers to the presence of more than one fetus in the current gestational period. Parental consanguinity was defined as consanguineous marriage between relatives in any degree.

Ethnicity was recognized by mothers, and ethnic group(s) among newborns’ ancestors as being: Latin Europeans, Non-Latin Europeans, Jewish, natives, Turkish, blacks, Asians, and others. These ethnic groups were later grouped according to complexion as whites, blacks, and brown-skinned subjects. Whites were those individuals whose African ancestry was totally discarded. Brown-skinned subjects were those who presented some kind of miscegenation between African race and any of the other races. We decided for the use of color self-concept and ethnicity as this is the approach employed by the Latin American Collaborative Study of Congenital Malformations, in spite of knowing that this determining factor is uncertain.

The presence of individuals with malformations in the family was attributed to ancestral congenital anomalies of any kind, according to parents, regardless of the degree of consanguinity with the newborn.

The data about the number of inhabitants of Pelotas were supplied by the Brazilian Institute of Geography and Statistics, based on the 1991 census, updated in 1996.

The sampling was obtained through the medical records of patients with malformations who participated in a large Latin American study (ECLAMC - Latin American Collaborative Study of Congenital Malformations), and was approved by the Science and Ethics Committees of all hospitals involved. The study did not require consent forms as it is a population study.

The study consisted of 81 cases and 4 control individuals for each case, allowing the detection (in 75% of the cases) of a 3.0 or higher relative risk, with significance of 5%.

Results

There were approximately 51,900 births in the maternity hospitals of the city of Pelotas throughout the study period (nine years). During this time, there were 81 cases of polydactylous newborns, thus determining a prevalence of 15.6 cases per 10,000 births.

Table 1 clearly shows the high prevalence of postaxial polydactylies, representing 73% of the cases registered in Pelotas. Hands were the predominant site of extra digit formation (82.5%), regardless of polydactyly type.

Most infants who were born with polydactyly were males (65.4%); the male/female ratio was 1.9:1. These differences, however, are not statistically significant (Table 2).

The variables weight at birth, mother’s age, twin pregnancy, parental consanguinity, number of pregnancies, and mother’s level of education (Table 2) did not show any significant difference between cases and controls.

The average birth weight for cases and controls was respectively 3,195g and 3,210g. The average age of mothers in both groups was 25 years. There was a birth of monozygotic twins among the study population, and both infants presented postaxial polydactyly of the hand. Five infants who also had twin brothers/sisters were among the

<table>
<thead>
<tr>
<th>Type of Polydactyly</th>
<th>Number of cases</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Postaxial</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hand</td>
<td>45</td>
<td>60.8</td>
</tr>
<tr>
<td>Foot</td>
<td>4</td>
<td>5.4</td>
</tr>
<tr>
<td>Hand and Foot</td>
<td>5</td>
<td>6.8</td>
</tr>
<tr>
<td>Preaxial</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hand</td>
<td>5</td>
<td>6.8</td>
</tr>
<tr>
<td>Foot</td>
<td>2</td>
<td>2.7</td>
</tr>
<tr>
<td>Unspecified</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hand</td>
<td>11</td>
<td>14.9</td>
</tr>
<tr>
<td>Foot</td>
<td>1</td>
<td>1.3</td>
</tr>
<tr>
<td>Hand and Foot</td>
<td>1</td>
<td>1.3</td>
</tr>
<tr>
<td>Total</td>
<td>74</td>
<td>100</td>
</tr>
</tbody>
</table>

Table 1 - Type and location of polydactylies. Pelotas, 1999
control individuals. Parental consanguinity was reported in 6.1% of the cases, and in 3.7% of control individuals.

Mothers of the cases and controls were very similar concerning their number of pregnancies, and level of education. In both groups, mothers reported an average of two pregnancies, including current gestation. Although 8.6% of cases were infants whose mothers had had more than six pregnancies against only 2.7% among control individuals, parity was not statistically significant between the groups.

Table 3 clearly shows the differences between cases and controls in terms of their ancestors’ complexion. When the study population was analyzed, adding their African and brown-skinned ancestors and white ancestors, we observed that infants who had at least one African ancestor represented 20.3% of the individuals in the control groups, and 45.8% of the cases. The existence of at least one African or brown-skinned ancestor was associated with general odds ratio 3.31 higher for polydactyly (OR (G)=3.31; 95%CI= 1.86-5.91; P<0.001).

The variable “family history of malformations” (Table 4) presented the greatest difference between cases and controls. Among the cases, 55% reported at least one relative with some kind of malformation. The percentage in the control groups was only 13% (OR (G)=10.18; 95%CI= 5.53-18.84; P<0.001). Only 81.3% of patients presented polydactyly while only 7.2% of ancestors who had some kind of malformation presented polydactyly among control individuals.

### Table 2 - Distribution and gross effects of maternal and newborn characteristics on the prevalence of polydactyly, according to case-control status. Pelotas, 1999

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Cases (n=81)</th>
<th>Controls (n=324)</th>
<th>OR(G) (95%CI)</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>53 (65.4%)</td>
<td>185 (57.8%)</td>
<td>1.38 (0.81-2.37)</td>
<td>0.27</td>
</tr>
<tr>
<td>Female</td>
<td>28 (34.6%)</td>
<td>135 (42.2%)</td>
<td>1.00</td>
<td></td>
</tr>
<tr>
<td>Twin pregnancy</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>2 (2.4%)</td>
<td>5 (1.5%)</td>
<td>1.62 (0.21-9.60)</td>
<td>0.64</td>
</tr>
<tr>
<td>No</td>
<td>79 (97.6%)</td>
<td>319 (98.5%)</td>
<td>1.00</td>
<td></td>
</tr>
<tr>
<td>Parental Consanguinity</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>4 (6.1%)</td>
<td>12 (3.7%)</td>
<td>1.92 (0.57-6.16)</td>
<td>0.22</td>
</tr>
<tr>
<td>No</td>
<td>62 (76.5%)</td>
<td>286 (88.2%)</td>
<td>1.00</td>
<td></td>
</tr>
<tr>
<td>Mother’s Age</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt; 20</td>
<td>14 (17.2%)</td>
<td>52 (16.0%)</td>
<td>1.00</td>
<td>0.80</td>
</tr>
<tr>
<td>20-25</td>
<td>26 (32.0%)</td>
<td>114 (35.1%)</td>
<td>0.85 (0.39-1.87)</td>
<td></td>
</tr>
<tr>
<td>26-31</td>
<td>21 (25.9%)</td>
<td>67 (20.6%)</td>
<td>1.16 (0.51-2.69)</td>
<td></td>
</tr>
<tr>
<td>&gt; 31</td>
<td>17 (20.9%)</td>
<td>80 (24.6%)</td>
<td>0.79 (0.33-1.87)</td>
<td></td>
</tr>
<tr>
<td>Number of Pregnancies</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1</td>
<td>27 (33.3%)</td>
<td>113 (34.8%)</td>
<td>1.00</td>
<td>0.39²</td>
</tr>
<tr>
<td>2</td>
<td>19 (23.4%)</td>
<td>84 (25.9%)</td>
<td>0.95 (0.47-1.90)</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>15 (18.5%)</td>
<td>51 (15.7%)</td>
<td>1.23 (0.57-2.65)</td>
<td></td>
</tr>
<tr>
<td>&gt;3</td>
<td>19 (23.3%)</td>
<td>63 (19.2%)</td>
<td>1.26 (0.62-2.57)</td>
<td></td>
</tr>
<tr>
<td>Mother’s Level of Education</td>
<td></td>
<td></td>
<td></td>
<td>0.39²</td>
</tr>
<tr>
<td>Low</td>
<td>57 (70.3%)</td>
<td>228 (70.3%)</td>
<td>1.00</td>
<td></td>
</tr>
<tr>
<td>Medium</td>
<td>14 (17.2%)</td>
<td>65 (20.0%)</td>
<td>0.86 (0.43-1.71)</td>
<td></td>
</tr>
<tr>
<td>High</td>
<td>3 (3.7%)</td>
<td>20 (6.1%)</td>
<td>0.60 (0.14-2.23)</td>
<td></td>
</tr>
</tbody>
</table>

(1) Different in the total of variables due to missing information. OR(G) (95%CI): General odds ratio and confidence interval of 95%.
(2) P for linear tendency.

### Table 3 - Distribution of ancestors of cases and controls according to complexion. Pelotas, 1999

<table>
<thead>
<tr>
<th>Complexion</th>
<th>Cases</th>
<th>%</th>
<th>Controls</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Black</td>
<td>16</td>
<td>22.2</td>
<td>33</td>
<td>11.2</td>
</tr>
<tr>
<td>Brown-skinned</td>
<td>17</td>
<td>23.6</td>
<td>27</td>
<td>9.1</td>
</tr>
<tr>
<td>White</td>
<td>39</td>
<td>54.2</td>
<td>235</td>
<td>79.7</td>
</tr>
<tr>
<td>Total</td>
<td>72</td>
<td>100</td>
<td>295</td>
<td>100</td>
</tr>
</tbody>
</table>
Table 4 - Family history of malformations according to case-control status. Pelotas, 1999

<table>
<thead>
<tr>
<th>Family history of malformations</th>
<th>Cases</th>
<th>%</th>
<th>Controls</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>45</td>
<td>55</td>
<td>42</td>
<td>13</td>
</tr>
<tr>
<td>No</td>
<td>28</td>
<td>35</td>
<td>266</td>
<td>82</td>
</tr>
<tr>
<td>Not informed</td>
<td>8</td>
<td>10</td>
<td>16</td>
<td>5</td>
</tr>
<tr>
<td>Total</td>
<td>81</td>
<td>100</td>
<td>324</td>
<td>100</td>
</tr>
</tbody>
</table>

OR(G) (95%CI): 10.18(5.53 - 18.84)  P<0.001

Discussion

In highly-miscegenated urban populations, different prevalence rates for congenital anomalies at birth largely reflect ethnic differences among countries. In South America, the prevalence of congenital anomalies is based on geographical differences, especially due to distinct proportions in the basic trihybrid “mix” of Amerindians, Latin Europeans, and African blacks.

In the city of Pelotas, the prevalence rates of congenital anomalies are mainly influenced by Latin European and African black origins. According to the 1991 census, updated in 1996, 85.5% of the local population are white, 7.43% black, and 7.2% brown-skinned (mulattos, mestizos, and other combinations of African, European and native Brazilian extractions). The remaining 0.32% is related to Asians or uninformed complexion, or individuals who were not able to inform their own skin color.

In Pelotas, isolated polydactyly is one of the most frequent malformations; only congenital club foot has a higher prevalence at birth.

The rate of 15.6 case per 10,000 births, obtained during the 9-year study period, predominantly corresponds to postaxial polydactyly. As the type of polydactyly was not specified in 20.9% of the cases, we believe that the prevalence of postaxial polydactyly of the hand is slightly higher. Therefore, the prevalence rates for preaxial polydactyly is around 1.7/10,000, and 13.9/10,000 for postaxial polydactyly.

Preaxial polydactyly may be associated with the Chinese and Amerindian ethnic groups, is nonfamilial, and usually affects just one side of upper limbs. A very small part of the population in Pelotas descends from the Chinese and Amerindians, which explains its low prevalence. Preaxial polydactyly is said to have an autosomal recessive form, and is usually involved in several syndromes; as a result, its genetic traits are related to these syndromes, shifting into autosomal dominant form, depending on the syndrome.

As to postaxial polydactyly, in most of the cases, authors such McKusick (1990) and also Frazier (1960) reported a prevalence 10 times higher in blacks than in whites. Scott-Emuakpor and Madueke (1976) found a total prevalence of 225/10,000 in Nigeria, whereas Castilla et al. (1997) reported a prevalence of 14.3/10,000 in Latin America, a prevalence rate that is very close to that found in Pelotas. Tenconi et al. (1990) reported a prevalence of 4.8/10,000 in Italy, and Handforth (1950) registered just one case among 5842 Chinese prisoners in Hong Kong. In this same study, 13 prisoners presented preaxial polydactyly.

In general, postaxial polydactyly is considered an autosomal dominant characteristic in humans, with low penetrance and variable expressivity, which also has an autosomal recessive characteristic and is associated with several syndromes. Orioli (1995) suggested segregation distortion of postaxial polydactyly due to the action of a recessive, gender-related modified gene, with higher incidence in blacks, especially when related to postaxial polydactyly of the hand. This genetic behavior is clearly present in the population of Pelotas affected by this malformation, in view of the high prevalence of postaxial polydactyly (especially of the hand) and the association of polydactyly with black ancestry, minority of local inhabitants, in addition to the large number of polydactyly.

This study did not reveal any significant difference as to gender, which can be explained through the high representativeness of boys among the control groups, due to the pair-up of these controls with newborns presenting other kinds of malformation. Even so, the male/female ratio was 1.9:1. Other authors reported a higher prevalence of this type of malformation in males. In previously mentioned studies, Scott-Emuakpor and Madueke (1976) reported a male/female ratio of 1.5:1 in Nigeria, the same as Tenconi et al. (1990) in Italy. In South America, Castilla et al. (1973) reported a male/female ratio of 1.3:1 for postaxial and 2.9:1 for preaxial polydactylies.

As to the postaxial polydactyly of the hand, Castilla et al. (1997) studied 1,733 cases in Latin America during 26 years, and reported an association of black African ancestry, males, twins, and low education level of mothers, and parental consanguinity, with frequent familial recurrence. The present study did not show any significant association between these characteristics and polydactyly. The sample size, however, had a power of only 75% to detect odd ratios higher than or equal to 3.0.

In this study, the occurrence of polydactyly was positively associated with the existence of black ancestry, and family history of congenital malformations. Black ancestry represented an odds ratio 3 times higher than that observed among white children. The most commonly characteristic associated with the presence of polydactyly was the positive history of malformations in other family members, with an
odds ratio 10 times higher than that observed in other cases. Polydactyly was the most frequently reported malformation among patients’ relatives (81.3%). The fact of having a family member with history of polydactyly resulted in an odds ratio 55 times higher for the presence of malformations in newborns (OR (G)=55.00; 95% CI= 26.02-118.25; P<0.001). Part of the results obtained in family history, especially in relation to polydactyly, originates from memory bias. Mothers of newborns with malformation recall facts and other previous malformations more frequently than mothers who gave birth to healthy infants. Mothers’ memory may have improved the total set of information (race, previous family history, etc.) used for comparison.

References