Acta Genet Med Gemellol 29:241–245 (1980)
The Mendel Institute/Alan R. Liss, Inc.

Case Report

Received 14 July 1980
Final 20 August 1980

A Discordant Case of Monozygotic Twins With Symbrachydactyly

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The case presented here may be the first report of symbrachydactyly in one child of monozygotic twins. Detailed testing of blood groups was performed to confirm zygosity. This case suggests that symbrachydactyly may be caused by intrauterine environmental factors.

Key words: Symbrachydactyly, Twins, Intrauterine factors

This is the first report, to our knowledge, of a discordant case of monozygotic (MZ) twins with symbrachydactyly.

CASE REPORT

Parents' History

The parents of the twins were not related. The mother married at 23 and was 25 years of age at the twins' birth. Her serologic test for syphilis was negative. The father was 28 years of age at the twins' birth. Both parents were healthy and of normal intelligence. No history of skeletal anomaly or mental defect was discovered in either side of the family. There also was no history of twinning on either side.

Obstetric History

The mother had no previous pregnancy. The presence of a twin pregnancy was confirmed by radiologic examination at the 28th week. There were no complications of pregnancy. There was no albuminuria, hypertension, or swelling of the ankles. Labor pain occurred spontaneously at the 36th week and male twins were born. The placenta was described as having a single membrane.

The Twins

Twin 1 weighed 2,208 g at birth and had asphyxia. He continued to be extremely cyanotic. Heart sounds were clear, rhythm was regular. He was diagnosed as having respiratory distress syndrome and received treatment for 32 days. Chromosome analysis was entirely normal. His left hand presented symbrachydactyly (Figs. 1 and 2). All fingers were reduced to rudiments.

Twin 2 weighed 2,203 g at birth and showed normal physical features. Physical development progressed normally (Fig. 3).

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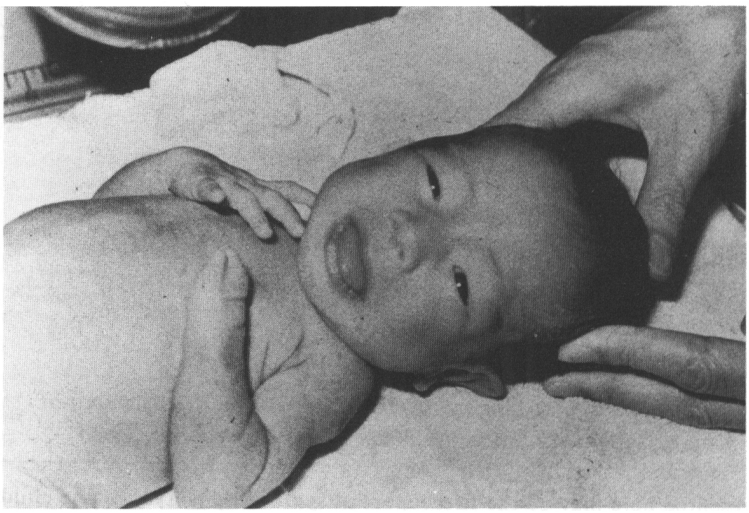

Fig. 1. Twin 1 with symbrachydactyly at 2 days of age.

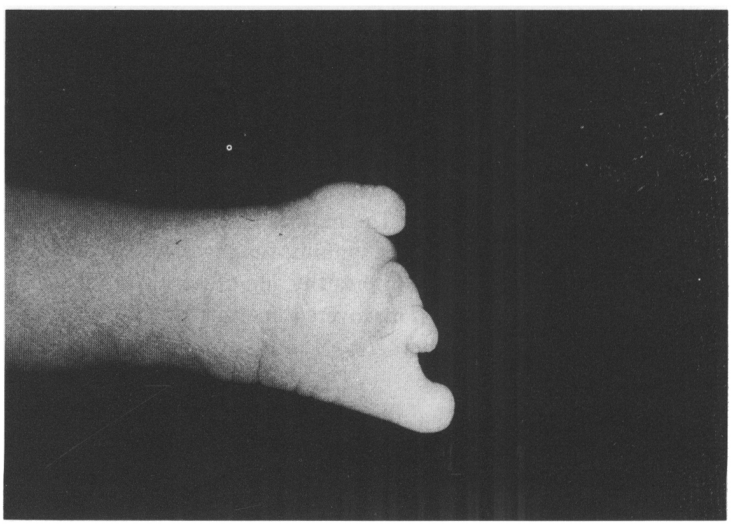

Fig. 2. Left hand of twin 1 with symbrachydactyly.

Dermatoglyphics

The dermatoglyphics of the twins are shown in Figure 4. In the symbrachydactylous hand, ridges run transversely across the palm and there is no axial triradius.

https://doi.org/10.1017/S0001566000007984 Published online by Cambridge University Press

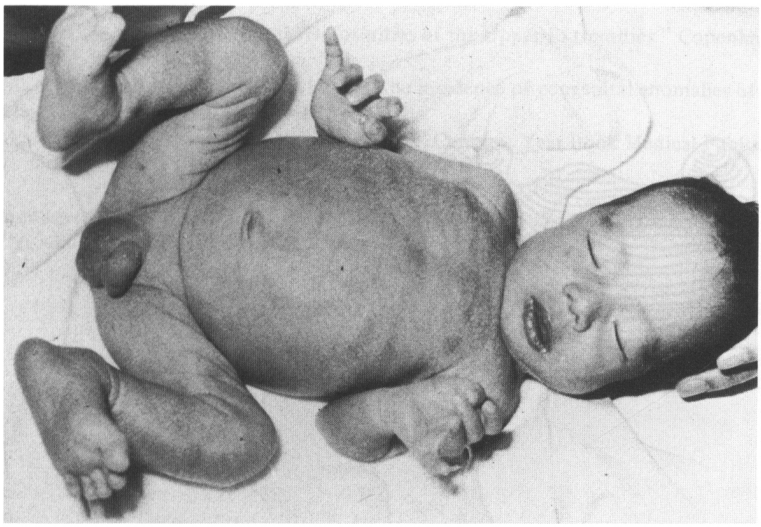

Fig. 3. Twin 2 at 2 days of age.

Zygosity

Detailed blood testing indicated all genetic markers examined to be identical in the twins (Table), so that they can be safely considered as MZ.

COMMENT

The chance of producing twins, one of whom has symbrachydactyly, is extremely small, and no case has, to our knowledge, been reported in the literature. The discordant occurrence of symbrachydactyly in MZ twins suggests that the pathology might be caused by intrauterine environmental factors.

The frequency of symbrachydactyly was about 1:50,000 in a Danish population [1] and about 1:44,000 in a Japanese population [2]. The frequency of twinning in Japan is approximately 1:144. Thus the probability of symbrachydactyly in twins is about 1 in 6.3×10^6.

There exist many forms and grades of symbrachydactyly. Hypoplasia affects the central rays more often than the ulnar and radial rays. In mild forms, there may be absence of the middle phalanges and underdevelopment of the distal phalanges of one or more of the central rays. In severe forms, the only remaining digit may be represented by the thumb, or all fingers may be reduced to rudiments [3]. In our case, all fingers are reduced to rudiments. In unilateral symbrachydactyly, there exist defects of the pectoralis muscle or chest wall, the so-called Poland syndrome. In this case, the defect of pectoralis major was not observed.

The discordant occurrence of symbrachydactyly in genetically identical twins suggests that this pathology is not genetic in origin, but rather due to intrauterine environmental factors.

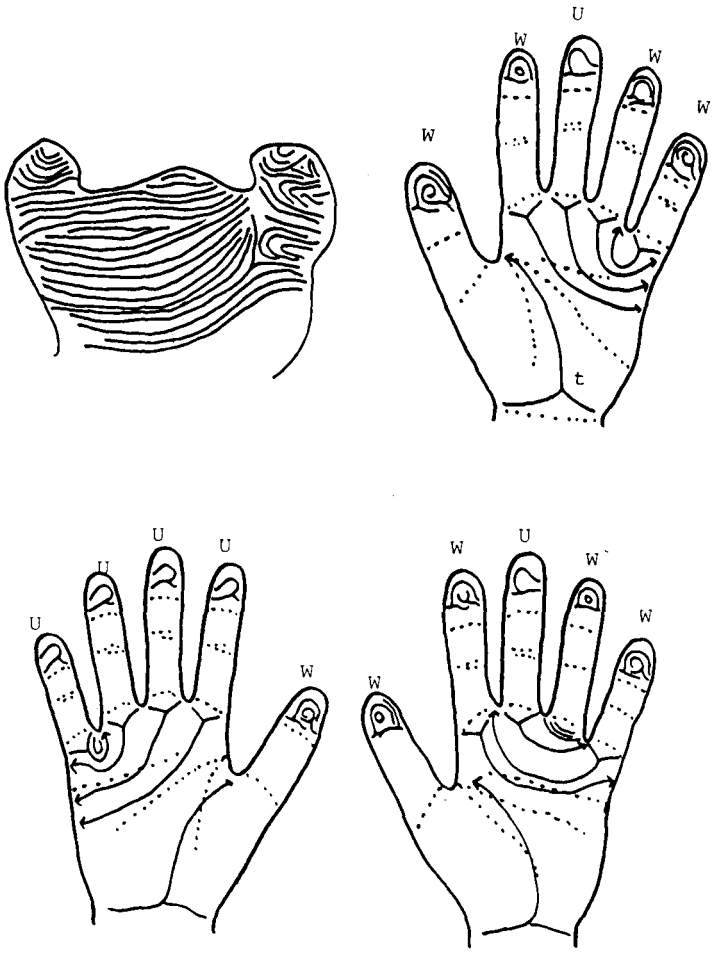

Fig. 4. Dermatoglyphic analysis of twins: Twin 1, top; twin 2, bottom.

TABLE. Blood Typing and HLA Typing of Twins

Twin 1	0.Nss.p.CCDee.Le(a−b−).Jk(a+).Lu(a−b+).Fy(a+b−)	
Twin 2	0.Nss.p.CCDee.Le(a−b−).Jk(a+).Lu(a−b+).Fy(a+b−)	
Twin 1	HLA Typing	A2,Bw22,B40,Cw3
Twin 2		A2,Bw22,B40,Cw3
Twin 1	Hp0−0	Gc2−1
Twin 2	Hp0−0	Gc2−1

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