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The Poland anomalad: a clinical and cytogenetic study of seven cases

By

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Seven patients with Poland anomalad, 6 males and one female, are reported. Four were affected on the right and 3 on the left side. The clinical, radiological and cytogenetic findings in these cases are discussed. In all the patients the nipples were asymmetric, the one on the affected side was sometimes set higher or lower than the one on the normal side, in relation to the extent of the muscle defect. One patient had coloboma of the iris. There was a greater variability of the defect of the upper extremity and especially of the ipsilateral hand than of the muscle anomaly. The hand seemed almost normal in 3 cases but these cases seem to represent the variability of the malformation. All cases were sporadic. Delivery was complicated in 4/7 and one patient had a twin brother with myelomeningocele.

The Poland anomalad or Poland syndrome [2, 3, 7] is the association of a thoracic malformation, essentially an aplasia or hypoplasia of the pectoralis major muscle, especially of its sterno-costal head, with brachysyndactyly of the ipsilateral hand. Absence of the pectoralis minor muscle, hypoplasia of the serratus magnus muscle, micromelia, nipple and mammary defects have also been described. Up to 1972, somewhat less than 100 cases have been published [6].

The anomalad is usually unilateral and occurs sporadically. The incidence amounts to about 1/30,000 births or between 1/10,000 and 1/100,000 [4]. The syndrome seems to show a great variability of expression. Mildly affected patients may have almost normal hands, while others are severely affected with dextrocardia [9] and renal anomalies [6, 10].

Recently, acute leukaemia has been reported [1, 5, 6] in patients with Poland anomalad. Accurate diagnosis is important to elucidate the relation that seems to exist between the syndrome and haematological malignancy.

To illustrate this point, we present here seven cases of the Poland anomalad and shall discuss the clinical manifestations, radiological and cytogenetic findings.

Case Reports

In all the patients, we have made a clinical study with measurements of the upper extremities and hands and also a radiological examination and comparison

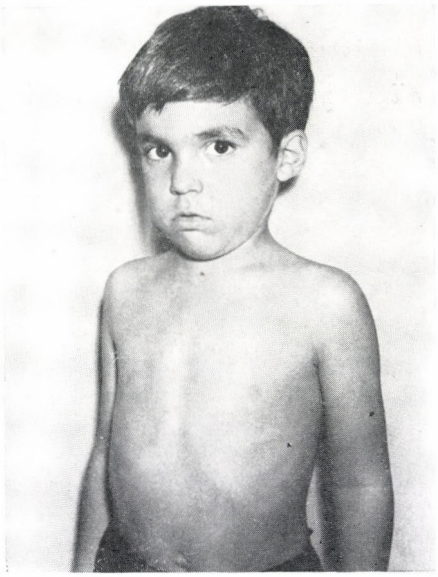

FIG. 1. Case 1. Poland anomalad on right side

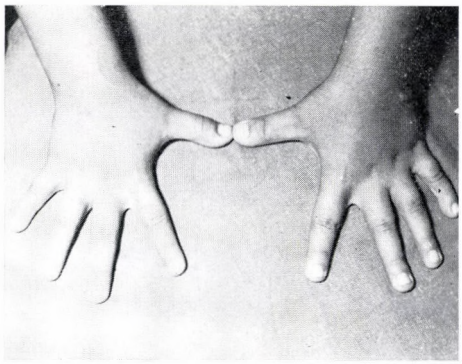

FIG. 2. The hands of Case 1: Slight webbing between fingers 2 and 3 on both sides

of the affected limb with the normal one.

Case 1. — J. A. M., a 3 8/12 year-old boy, was born 2.6.1971, after an eight-month gemellary pregnancy. The father and the mother were 27 years old at the time of the patient's birth. There was no consanguinity. Birth weight was 2.8 kg. There was no history of maternal drug habit, viral infection or X-ray exposure during pregnancy. The mother delivered twin boys by Caesarean section. The twin brother of the propositus had myelomeningocele and died 6 days after birth. The mother had had no spontaneous abortion, but in a previous pregnancy had delivered a boy who died 3 days after birth, with dextrocardia and a single ventricle. The patient is the only child. The mother during pregnancy had had pyelonephritis and received sulphonamides, mandelamide and nitrofurantoin for more than one month. She also had a positive glucose tolerance test after the

fifth month of gestation. There was no history of diabetes in her family. A chest X-ray was made in the 7th week of pregnancy. The husband had a pararectal fistula which dated from his youth, and his father had also had one in his youth.

The child's general health had been excellent. Mental and motor development were normal. There was no history of respiratory or urinary infection. At physical examination his height was 99 cm, weight 20 kg, head circumference 52 cm. He had a slight defect of the right helix with normally set ears, mild synophrys, malocclusion of the teeth, high arched palate, short neck, hypoplasia of the right pectoralis major muscle, especially its sternocostal head, right hypoplastic nipple set higher than the left one (Fig. 1). He had a funnel chest with moderate prominence of the chondrocostal junction. Both hands appeared to be normal, but there was a slight web between fingers 2 and 3 of both hands (Fig. 2). On the right palm, there was an aberrant Sydney line, and clinodactyly of the left 5th finger. The child is lefthanded. There was no dectrocardia. Four "café au lait" spots were found on the left arm. Intelligence was normal. Laboratory data included a normal haemoglobin concentration, haematocrit and white-cell count. X-rays of the thorax, upper extremities, spine and skull showed no alterations.

Case 2. — J. N. C., a 19-month-old boy, was born 29.9.1974. The father and mother were unrelated and were 47 and 44 years old respectively. The pregnancy was uneventful. This was the mother's fourth pregnancy. She took no drugs and had no viral infection or exposure to radiation during pregnancy.

The child's birth weight was 3.4 kg. His 3 brothers, 30, 27 and 26 years old, respectively, are normal. There was no family history of congenital malformations. The propositus had had no respiratory or urinary infections. At the age of 18 months he had a throat infection with fever and generalized convulsions of five minutes duration. Physical examina-

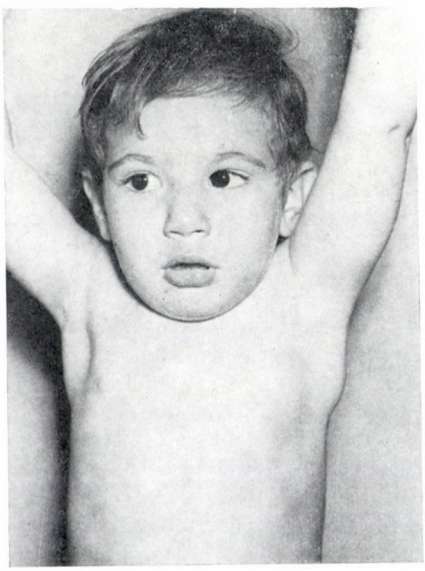

FIG. 3. Case 2. Defect of left pectoralis major muscle, hypoplasic left nipple lying lower than the right one

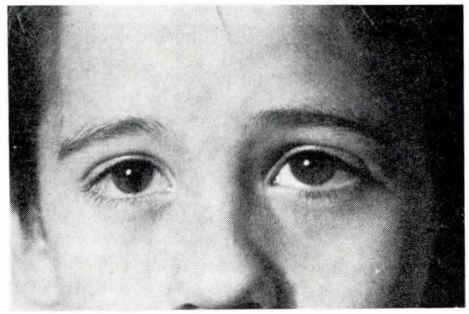

FIG. 4. Case 3. Coloboma of right iris

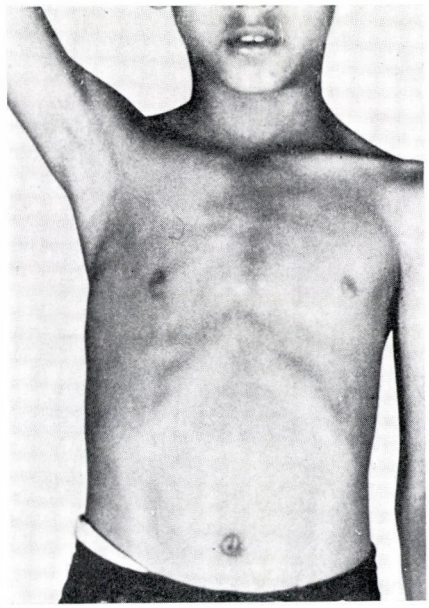

FIG. 5. Case 3. Poland anomalad on right side

tion data: height: 83 cm, weight: 14 kg, head circumference: 50 cm. There was no facial asymmetry. His neck was short and broad. The left pectoralis major muscle was almost absent. The left nipple was hypoplastic and set lower than the right one (Fig. 3). Café au lait spots were present on the anterior surface of the right hemithorax. The upper extremities were equal in size and length. The hands were normal, there was no simian crease or Sydney line on the palms. The boy had flat feet. Laboratory data included a normal haemoglobin concentration, haematocrit and white-cell count. X-rays of the skull, spine and upper extremities and thorax showed no alterations.

Case 3. — C. A. A., a 9 1/2 year-old boy, was born at term (6-5-1966), after a normal delivery. The mother was 22 and the father 23 years old. There was no consanguinity. The child weighed 3.4 kg at birth. The mother had noted an exanthematous rash in the fifth month of preg-

nancy. There was no history of parental irradiation and the mother had no medication other than vitamins. This was her 2nd child. The 12-year-old brother is normal. The mother had 4 brothers and 5 sisters with no congenital defects.

The patient's mental and motor development had been normal. He had had pneumonia on four occasions and pyelonephritis when he was 8 1/2 years old. On physical examination, his weight was 25 kg, height 133 cm, head circumference 51 cm. Malposition of the teeth and a high arched palate were observed, as well as coloboma of the right iris (Fig. 4). He had a rather long neck. The right costal head of the pectoralis major muscle was hypoplastic in its lower part. The right nipple was set lower than the left one. There was no axillary pterygium (Fig. 5) or dextrocardia. Three café au lait spots were present, one on the right cheek, one on the chin and one on the right arm. Both upper extremities were of the same size and length, but the right hand was smaller. There was increased soft tissue webbing between all the fingers of both hands (Fig. 6), with hyperextensibility of both thumbs and a simian crease on the right palm. The boy was left-handed. There was a wide gap between the first and second toes on both feet. Laboratory data showed normal blood counts, haematocrit and haemoglobin concentration, and urine analysis. Roentgenograms of the skull, chest and spine were normal. X-rays of the hands showed membraneous syndactyly between all the fingers on both hands. The right hand was smaller, with brachydactyly and less developed carpal bones. There was also a smaller interdigital space between the right thumb and second finger. The distal phalanx of the right thumb was puntiform.

Case 4. — D. G. G., a 3-year-old girl, was born 24-8-1973. At that time, the father and mother were 36 and 41 years old, respectively. There was no consanguinity. The child was born by Caesarean section, after a normal nine months pregnancy. Her birth weight was 3.2 kg. The mother had a fall in the 5th month of gestation and denied having had viral infection or having received any medication. The mother's third pregnancy ended in a spontaneous abortion. The child had two half sisters, 27 and 20 years old. The mother is the eldest of a sibship of seven. She had four brothers and two sisters. Two brothers died, and one, 36 years old, was born with malformed feet but it was not possible to examine him.

The patient had always been in good health and her intelligence is normal. On examination, weight and height were 25 kg and 91 cm, respectively, the head circumference was 48 cm. She had normally set ears with underdeveloped lobes. Her neck was short. The sternal head of the left pectoralis major muscle was absent. She had a moderate pectus excavatum in the lower third of the sternum. There was

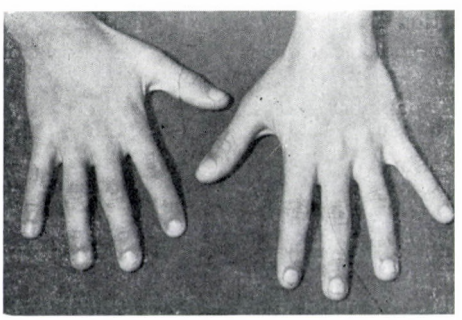

FIG. 6. The hands of Case 3. The right hand is smaller and there is a membraneous webbing between all the fingers

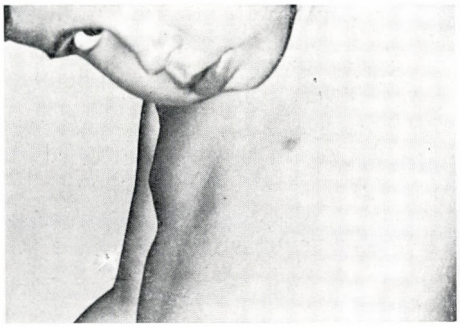

FIG. 7. Case 4. Poland anomalad on left side

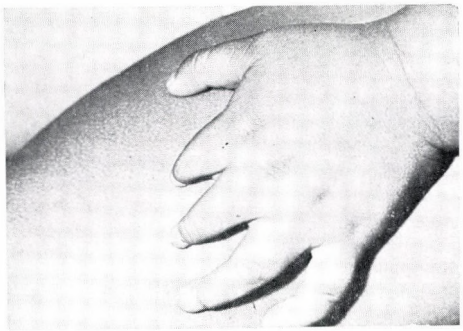

FIG. 8. Left hand of Case 4. Brachydactylic fingers and triangular shape of digits 2 and 3

asymmetry of the nipples, the left being higher than the right (Fig. 7). Both upper extremities were equal in size and length but the left hand was smaller. There was brachydactyly of all the left fingers, more marked in the 2nd and 3rd fingers. These digits had a triangular shape (Fig. 8) and the nails of the first, second and third left fingers were hypoplastic. Both thumbs were hyperextensible, clinodactyly of the fifth left finger was present as well as a simian crease on the left palm. No café au lait spots were seen. Laboratory data showed normal haemoglobin concentration, haematocrit, white-cell count and platelet count. X-rays of the chest, skull and upper extremities were normal. Roentgenograms of the hands showed that the left hand was smaller, with short 2nd and 3rd left metacarpal bones and absence of the intermediate and distal phalanges of the same fingers. There was also brachydactyly of the left fingers with clinodactyly of the fifth. The first three fingers of the left hand had a triangular shape.

Case 5. — P. L. P., a 15-year-old boy, was born at term (22-1-1961). Maternal and paternal ages were at that time 21 years. After a fall in the fifth month of gestation, the mother had an incipient abortion. She also had cystitis. No history of drug ingestion, viral infection or X-ray exposure were mentioned. After an abnormal delivery complicated by placenta previa, the child was born with 3.1 kg.

The propositus is the oldest son; he has two healthy brothers, 14 and 12 years old. The parents are not related and the mother had had no spontaneous abortion. The patient's health had always been good but recently he had hepatitis. His intelligence is normal. On examination, his weight

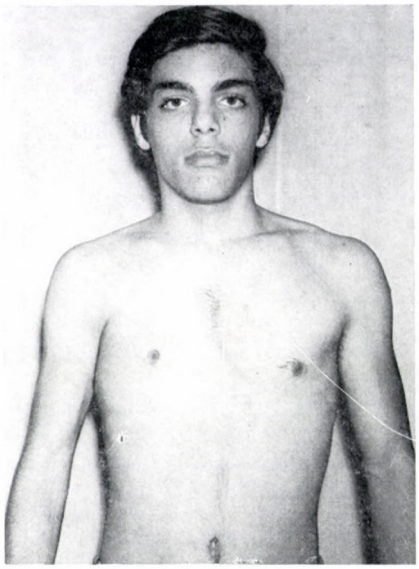

FIG. 9. Case 5. Hypoplasia of right pectoralis major muscle, the right nipple lies higher than the left one

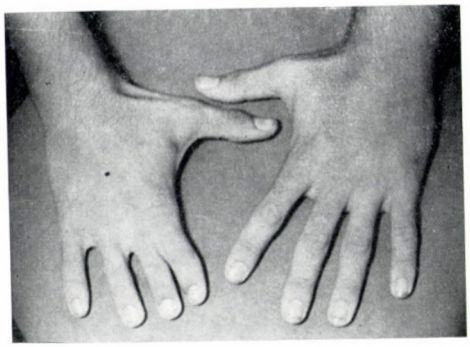

FIG. 10. The hands of Case 5. The right hand is smaller and there is brachysyndactyly

was 60 kg, height 164 cm and head circumference 56 cm. He had a slight synophris with moderate antimongoloid slant and malocclusion of the teeth. The costal head of the right pectoralis major muscle was absent with the ipsilateral nipple set higher than the left one (Fig. 9). Dorso-lumbar scoliosis was also present. The upper extremities were of the same length but the right hand was smaller in size. There was brachysyndactyly of the right hand (Fig. 10); the syndactly was more marked between the 2nd and 3rd fingers but was also present between the 3rd and 4th ones. There were no café au lait spots. Laboratory data revealed a normal haematocrit, haemoglobin concentration and white-cell count. Roentgenograms of the chest and skull were normal. X-rays of the upper extremities showed that the humeri were equal in length but the right internal condylus

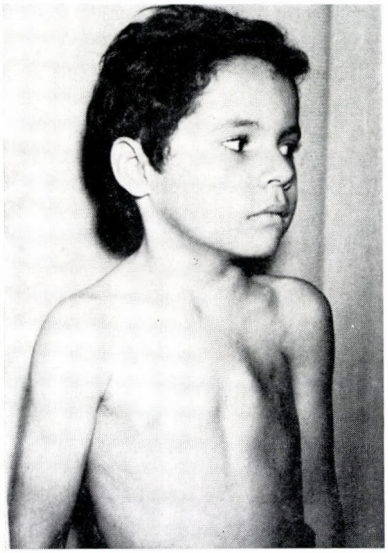

FIG. 11. Case 6. Poland anomalad on left side. Prominence of the chrondrocostal junction

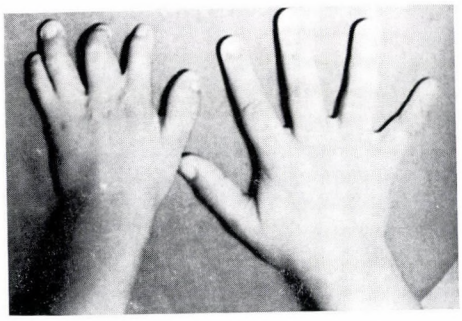

FIG. 12. The hands of Case 6. Brachydactyly with syndactyly on left side

was less developed. Radius and ulna were of the same length on both sides. There was brachysyndactyly with short metacarpal bones and the thumb was smaller than normal. There was also a dorso-lumbar scoliosis with bifid spine (S1).

Case 6. — G. T. G., a 7-year-old boy, was born 10-5-1969, to a 21-year-old mother and a 24-year-old father, after a complicated pregnancy during which two doses of progesterone were administered to the mother. At 6 $^1/_2$ months of pregnancy, the mother was hospitalized for incipient premature labour with rupture of membranes. This was her first pregnancy. There is no consanguinity or congenital malformation in the family. The child had had asthma during infancy and pneumonia on one occasion. Two surgical interventions were made to repair the syndactyly.

On examination, the propositus measured 122 cm and weighed 25 kg, his head circumference was 53.5 cm. He had a moderate synophrys, a slight retrognathism, the teeth were small and separated and the neck was short. The left pectoralis major muscle was hypoplastic with asymmetry of the nipples, the left being higher

than the right (Fig. 11). He had a funnel chest with prominence of the costochondral junction on the left side. The left upper extremity and the ipsilateral hand were smaller. The left fingers were shorter and the 3rd and 4th were flexed (Fig. 12). A membraneous syndactyly was present between the 2nd and 3rd, the 3rd and 4th and the 4th and 5th fingers of the hand. No nail was present on the 5th finger. The left thumb was broad and short with a hypoplastic nail and there was a simian crease on the left palm. There were surgical scars between fingers 2—3 and 3—4. No café au lait spots were detected. The laboratory data were normal and so were the chest and skull X-rays. The left metacarpal bones were shorter than the right ones and the middle phalanges of the 2nd and 4th fingers were lacking, with a rudimentary phalanx on the 3rd and 5th fingers. Distal phalanges were absent also on fingers 3—4—5. There was a puntiform distal phalanx on the left 1st and 2nd fingers and membraneous syndactyly between fingers 2—3—4—5 with radial deviation of the distal phalanx of finger 2. The left hand was undersized. The radius and ulna showed no alterations on either side.

Case 7. — O. M. A., a 4-year-old boy, was born 8-9-1972. The mother then was 26 years old and the father 35 years old. They were not related. The child was born at term after a normal delivery, with 3.4 kg. There is no history of the mother having taken drugs, having had viral infection or having been exposed to radiation during gestation. This was her 2nd pregnancy; the first ended with an artificial abortion. The child's health had been good, at examination his weight was 18 kg, the height 102.5 cm, head circumference 51.5 cm. He had small separated teeth, a short neck, the right pectoralis major muscle was lacking, the right nipple was set lower than the left one (Fig. 13). The right upper extremity was underdeveloped; length and size of the arm and forearm were smaller than normal. The right hand was hypoplastic with hyperextension of the wrist, flexed fingers and adducted thumb (Fig. 14). The thenar and hypothenar eminences were flat, a complete simian

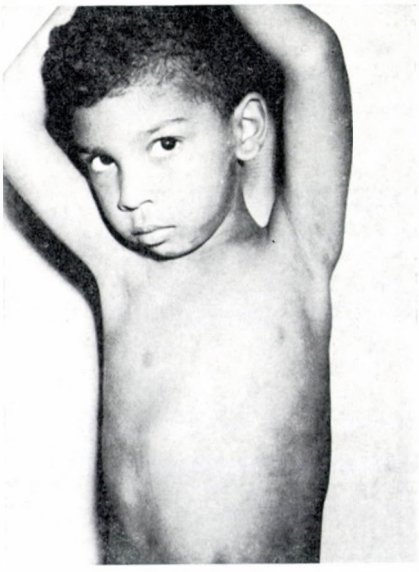

FIG. 13. Case 7. Hypoplasia of right pectoralis major muscle

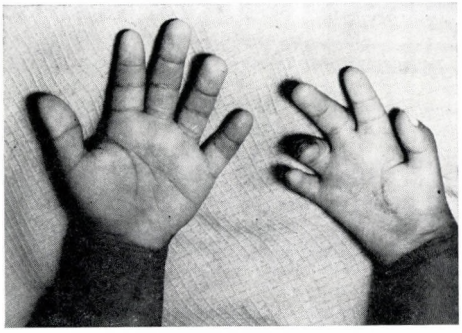

FIG. 14. The hands of Case 7. The affected hand is smaller with flexed brachysyndactylic fingers

crease was present on the right palm and an aberrant one on the left. The 4th and 5th right fingers were contracted. All the fingers were brachysyndactylic and a membraneous syndactyly was present between all fingers except between the thumb and the 2nd digit. He was left handed. Haemoglobin concentration, haematocrit and white-cell count were normal. Roentgenograms of the upper extremities showed that the ulna, radius and humerus on the right side were shorter. There was an ulnar deviation of the right fingers with shorter metacarpal bones, membraneous syndactyly, a shorter first interdigital space and osteoporosis. X-rays of the chest and skull were normal.

Cytogenetic Study

A buccal smear for sex chromatin yielded normal results in all the cases. Examination of metaphases from peripheral blood cultures revealed a normal set of 46 chromosomes in 5 cases. In two cases, banding of the chromosomes by trypsin solution and Giemsa staining showed no alteration.

Discussion

Six of the patients were males and one was a female. According to Mace et al. [6] the Poland anomalad shows a male predominance of 3 : 1 and the right side is affected in about 75% of the cases. In our series the defect was on the right side in 4 and on the left in 3 children. The anomaly of the pectoralis major muscle was marked in 3 (Cases 2, 5 and 6) and slight in 4 cases. Three children had pectus excavatum. A few small café au lait spots were present in 3 children. Asymmetry of the nipples was seen in all the cases. We have observed in our series and in the pictures of other reported cases that the nipple on the affected side was sometimes set higher and sometimes lower than the one on the normal side. This sign might be related to the extent of the pectoralis major muscle defect.

None of our patients had rib defects, herniation of the lungs or dextrocardia.

There was a greater variability of the defect of the ipsilateral upper extremity. Arm length was the least variable, being normal in 5/7. Its width was smaller in 3/7. The forearm was shorter in one but its width was less in 3/7. In one case, the hand seemed normal but was small and there was a slight membraneous webbing between the fingers, which were short. This patient had also some bone alterations on X-ray examination. Another child had slight webbing between two fingers, a Sydney line on the palm and clinodactyly of the 5th finger. Case 2 had a normal hand and a normal osseous structure of the upper extremity. Brachydactyly was seen in 5/7 with syndactyly in 4/7 and without syndactyly in one. There was syndactyly alone in one case. In two cases, a flexion contracture of the fingers was present. Case 7 had a malformed hand similar to that of the case reported by David and Saad [3]. As to the brachydactyly, in Cases 4, 5, 6 and 7, the metacarpal bones were short and in Cases 4 and 6 the intermediate phalanges were missing. The distal phalanges were absent in two children. The syndactyly was slight in Cases 1, 3 and 7 and was present between all the fingers except between thumb and index in Cases 3 and 7. In Cases 5 and 6, the syndactyly was more marked and in Case 5 only between the 2nd and 3rd fingers. The patients of Mace et al. [6] showed brachydactyly on the side of the muscle defect in 4/7, absence of phalanges or digits in 3/7. One of their patients had no thumb, and syndactyly was seen in 4/7. Variability was marked in the anomaly of the hand and the degree of the hand defect did not seem to be closely related to the extent of the pectoralis major anomaly. The slight malformation of the hand may be overlooked, as we showed in 2 of our patients, and it seems that in cases called unilateral malformation of the pectoralis major muscle, the hand defect may have been undetected if it was moderate. One of our patients had a malformation not reported until now, a coloboma of the iris on the same side as the muscle defect.

No attempt at abortion was mentioned in our series, in contrast to that of David [2]. Delivery was complicated in 4, and placenta previa and ruptured membranes occurred in 2 of the mothers. In Case 1, the twins were of the same sex and both had congenital defects: one had Poland anomalad and the twin who died, a myelomeningocele. Myelomeningocele is a closure defect compatible with a multifactorial/threshold explanation and this might be a possibility also as regards the Poland anomalad.

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