DISCORDANT DEFORMITIES OF LIP AND PALATE IN MONOZYGOTIC TWINS A REPORT OF 2 SETS OF TWINS

By Khoo Boo-Chai

Discordant deformities of the lip and palate in monozygotic twins are rare. Since the twins arise from the same egg cell after fertilisation by a single spermatozoon, we would expect the same condition to be duplicated in the other twin. The congenital abnormality occuring in one twin only, or in both, but differing in severity and location is extremely interesting from an aetiologic standpoint because it shows that in some cases, non-genetic factors are operative during the formation of the lip and palate. The author came across two sets of such cases and because of their rarity, decided to have them documented.

1ST SET OF TWINS

ABE, a Chinese girl aged 17 years, was first seen by me on February 2, 1964. She came for a secondary revision of her cleft lip and nose. She had a primary repair elsewhere when she was 7 months old but the results were not to her entire satisfaction. A straight line closure was used and on closer physical examination (Fig. 2) the following abnormalities were noted:-

(i) Marked scarring of the upper lip which was tight from side-to-side.

(ii) A right oro-nasal fistula which was 4 mm. in diameter.

(iii) Marked asymmetry of the nostrils with drooping of the right alar margin.

(iv) A whistle deformity of the upper lip.

(v) A marked irregularity of the vermillion of the upper lip.

(vi) An unrepaired cleft palate on the right side involving the whole of the soft palate and the hard palate.

On further questioning, she said that she had an elder twin sister (ABL) who was normal (Figs. 1A & B). Her mother volunteered the information that there was only "one placenta" and this was told her by the attending midwife. Her two other brothers were normal. There were no other members in the family with this deformity. It is interesting to note that of the families followed-up in our series, 15% of them had more than one member with clefts. (Khoo Boo-Chai, 1965).

Figure 1 A.

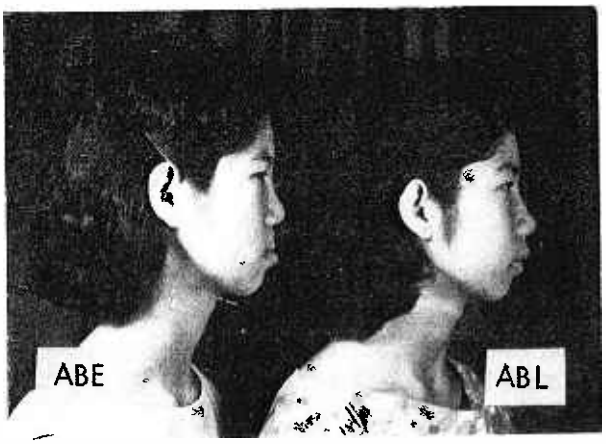

Figure 1 B.

Figs. 1A & 1B. The first pair of monozygotic twins. The one on the left has a complete cleft lip which was repaired and an unrepaired cleft palate. The other twin is normal.

It was obviously of some importance to establish conclusively the zygosity of this pair of twins because only one had the deformity. There was a strong physical resemblance to one

another (Fig. 1) and they were of the same blood group. The blood groups were typed as follows:-

ABE (Patient)

Anti — A ... ... Positive
Anti — Al ... ... Positive
Anti — B ... ... Negative

ABO Group Al

Anti — M ... ... Positive
Anti — N ... ... Positive

MN Group MN

Anti — C (rh') ... Positive
Anti — D (Rho) ... Positive
Anti — E (rh") ... Positive
Anti — c (hr') ... Positive
Anti — e (hr") Positive
Most probable genotype CDe/cDE.

Cross Match

No agglutination with cells to plasma of ABL and plasma to cells of ABL.

ABL (Twin sister)

Anti — A ... ... Positive
Anti — Al ... ... Positive
Anti — B ... ... Negative

ABO Group Al

Anti — M ... ... Positive
Anti — N ... ... Positive

MN Group MN

Anti — C (rh') ... Positive
Anti — D (Rho) ... Positive
Anti — E (rh") ... Positive
Anti — c (hr') ... Positive
Anti — e (hr") ... Positive
Most probable genotype CDe/cDE.

Cross Match

No agglutination with cells to plasma of ABE and plasma to cells of ABE.

18.11.64: A reciprocal skin homograft was performed under local anaesthesia. A full thickness skin graft 20 mm. in diameter taken from the medial aspect of the left upper arm near the axilla was exchanged. The grafts took and are still viable at the time of writing which is more than one year after operation (Fig. 3). Human skin homografts exchanged between dizygotic twins are usually rejected from the nineteenth to the twenty-ninth day.

Figure 2 A. Figure 2 B.

Fig. 2A. A close-up photograph showing the straight line closure with distortion of the configuration of the upper lip.

Fig. 2B. Photograph taken 6 months after revision of the lip and nose.

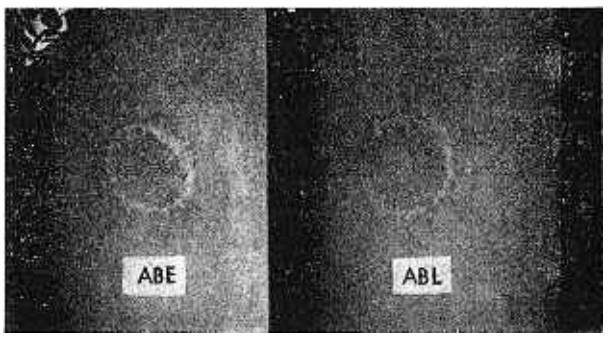

Fig. 3. Photograph taken one year after a reciprocal skin homograft operation. A full thickness skin graft was exchanged from the medial surface of the left upper arm.

10.2.65: A secondary repair of the cleft lip with revision of nose was performed under local anaesthesia with heavy sedation. The square flap technique (Trusler-Glanz) was used for the lip repair. Since there was a discrepancy in the vertical height of the lip, a wedge was removed from under the right alar base. The deformed nose was treated like that of a bifid nose (Khoo Boo-Chai, 1965). The two alar cartilages were

dissected free from their skin cover and properly positioned with nylon sutures. The nasal floor was repaired with contiguous skin flaps.

2ND SET OF TWINS

Both WTK and WTN were first seen by me on January 14, 1966. The mother actually brought the elder twin WTK for an opinion regarding lip repair. He had a right sided cleft of the lip and palate (Figs. 5A & 5B). WTN tagged along because there was no one else to look after him in the house. He also had a deformity of lip on the left side which was detected only on close physical examination (Fig. 5C).

1st Twin: WTK.

Birth weight : 5 lbs. 14 ozs. Sex: Male.
Present weight : 17 lbs.
Deformity : **Right side:** A cleft of the lip and palate.

2nd Twin: WTN.

Birth weight : 8 lbs. 0 oz. Sex: Male.
Present weight : 19 lbs.
Deformity : **Left side:** The mildest form of unilateral cleft of the lip with notching of the vermillion. unequal nostrils and a minor degree of subcutaneous muscle defect.

There was a marked physical resemblance between the two (Fig. 4). The mother was told by her attending obstetrician that there was only one placenta. There was no history of any bacterial or viral infection or of dietary deficiency in the first trimester of pregnancy. Apart from the cleft of the lip and palate, no other congenital abnormalities were found.

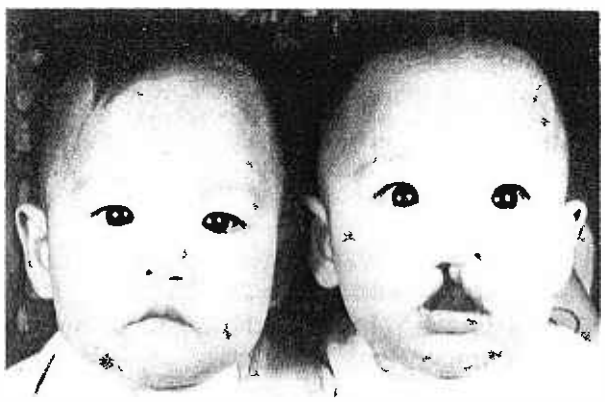

Fig. 4. The second pair of monozygotic twins. The first twin has a cleft of the lip and palate on the right side. The second twin has a mimimal cleft of the lip on the left side.

Fig. 5A. A close-up photograph of the first twin.

Figure 5 B.

Figure 5 C.

Fig. 5B. Photograph taken 2 months after surgery using the triangular flap technique. Note the normal pout of the lip together with the preserved Cupid's bow. Also note the symmetrical nostrils.

Fig. 5C. A close-up photograph of the second twin. Note the asymmetrical nostrils, the vertical groove on the upper lip and the notched vermillion.

It is interesting to note that the earliest case of a successful cleft lip repair was recorded over a thousand years ago in ancient China, circa AD 390. (Khoo Boo-Chai, 1966). However, it is only during the last two decades that there have been rapid improvements in the technique of the lip repair.

Fig. 6. Photograph taken 2 months after reciprocal skin grafting. A full thickness skin graft 20 mm in diameter was exchanged from the medial surface of the right upper arm.

A primary repair of the unilateral cleft lip using the triangular flap technique (Khoo Boo-Chai, 1965) was done on WTK on January 23, 1966 under general anaesthesia. Towards the end of the operation, the second twin WTN was anaesthetised and a reciprocal skin graft operation performed. A full thickness skin graft 20 mm in diameter was taken from the medial aspect of the right upper arm and exchanged with a similar graft from the first twin. The grafts took and are still viable at the time of writing (Fig. 6).

DISCUSSION

The cleft lip and/or palate deformity may occur only in one monozygotic twin (Rogers, 1957; Metrakos et al, 1958) or, they may occur in both but the side and severity may differ. (Ramsey and Wynn-Williams, 1960: Peer, 1958). This is interesting because it indicates that in some cases, there are factors other than genetic ones that cause the formation of clefts. These factors must operate before the 12th week of intra-uterine life because the primary palate forms before the 7th week and the secondary palate before the 12th week. This observation has led some investigators to give big doses of vitamins to pregnant mothers who have borne ftcle lip and/or palate babies previously. The results, so far, have been inconclusive.

The other point of interest lies in the use of skin homografts in twins. Therapeutically skin homografts are used for skin replacement in burns in one of the twins. (Converse and Duchet, 1947). In certain medico-legal cases, reciprocal skin grafting has been most helpful. (McIndoe and Franceschatti, 1950). In our cases it is used to determine the zygosity of the twins. Recently, Dencker and associates (1961) have shown that among dizygotic twins of the same sex, dizygosity was proved in about 95% of the twin pairs if most or all the currently known blood group systems were used. There is still 5% of dizygotic twins with identical blood groups. Hence, this line of investigation has its limitations. The history of one or two placentae is again not entirely reliable especially from our local mothers. The presence of two placentae is not always indicative of dizygosity. It is only when there is a mono-chorionic placenta (mono—or diamniotic) that the monozygosity of the twins is certain. Examination of the physical characteristics and the use of finger prints may help but they are not conclusive in difficult cases. To Rogers (1955) must go the credit for a careful and complete study of the behaviour of skin homografts in human twins. He has shown that reciprocal skin homografting, can be employed to diagnose human monozygosity or dizygosity for purposes of determining the aetiology of hereditary and congenitally acquired abnormalities and in certain cases presenting medico-legal quandries. However, there are 2 reservations—in the very rare condition of twin human chimeras and in agammaglobulinaemia one must be aware that the rejection phenomenen theoretically may not occur.

SUMMARY

Two sets of monozygotic twins with discordant deformities of the lip and palate are recorded. The zygosity of these cases was confirmed by reciprocal skin grafting.

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