Acta Genet Med Gemellol 34:185-188 (1985)


Received 2 May 1985

*CASE REPORT*

# Discordant Monozygotic Twins with Trisomy 13

**H.T. Loevy, Marilyn Miller, Ira M. Rosenthal**

*Departments of Pediatric Dentistry, Ophthalmology and Pediatrics, University of Illinois at Chicago*

**Abstract.** Monozygotic twins with typical trisomy 13 are reported. Despite an identical karyotype, the twins were dimorphic for the presence of an omphalocele. Reasons for the rarity of MZ twins with trisomy 13 are presented. It is suggested that the presence of a chromosomal abnormality in MZ twins may predispose to dimorphism.

**Key words: Monozygotic twins, Trisomy 13, Discordant, Omphalocele**

There have been a number of cases of MZ twins with trisomy 21 [1,2,5,8-11,15,16,18-22]. There are also reports of MZ twins with 45,X [12], 48,XXY,+21 [5], trisomy 8 with mosaicism [17], and ring 18 trisomy with mosaicism [3]. Significant dimorphism has been reported in some of these twins [3,4,8,9,11,16,17,20,22].

We are reporting MZ twins with trisomy 13. Despite identical karyotypes, dimorphism was manifested by the presence of an omphalocele in one of the twins.

## REPORT OF THE CASES

Male twins were delivered at term by Cesarian section. Placental examination showed one chorion and two amniotic sacs. Because of multiple congenital anomalies, the twins were transferred shortly after birth to the University of Illinois Hospital. The mother, gravida 4, was 23 years old. There had been 3 previous elective abortions. No abnormalities were detected in this pregnancy, and there was no history of chromosomal abnormalities or congenital malformations in the family.

**Twin 1** weighed 2800 g. Bilateral cleft lip, cleft palate, low set ears, and microcephaly were present (Fig. 1). There was no polydactyly. The palpebral fissures were 5 mm in horizontal diameter with incomplete separation of the eyelids.There was an entropion of each upper lid. Bilateral microphthalmus was present, the corneal diameter measuring 5 mm in each eye. Presence of bilateral corneal leukomas precluded further evaluation of the anterior and posterior segments of each eye.

Dextrocardia was present with a grade II of VI systolic murmur. An ECG at the age of 2 days revealed a heart rate of 120/min with sinus arrhythmia; PR was 0.12. There were deep Q waves in

leads I, II and aVF and a QS pattern in aVL. Upright T waves were present in V4r and V7. The prominent Q waves suggested septal hypertrophy or positional abnormality. Chest roentgenogram confirmed the presence of dextrocardia.

An EEG showed sharp waves in the frontal and temporal areas indicating the presence of discharging foci.

Blood type was A+. A karyotype, prepared from peripheral blood lymphocytes, revealed a 47,XY,+13 pattern. After the parents were informed of the karyotype and its significance they refused further diagnostic studies.

The infant suffered from numerous apneic spells and died suddenly at the age of 19 days. Consent for autopsy was refused.

**Twin 2** weighed 1900 g and had an appearance similar to twin 1 with microphthalmia, bilateral cleft lip, cleft palate, low set ears and microcephaly (Fig. 2). There was no polydactyly. Eye findings were similar to twin 1. The fissures were small, measuring 7 mm in the right eye with incomplete closure of the nasal sector. Vertical diameter was approximately 2 mm. The left fissure was narrower in horizontal and vertical dimensions. A downward slant of both fissures was present. Bilateral corneal leukomas were present, preventing further evaluation of the anterior and posterior segments.

Dextrocardia was present. There was a grade I of VI soft systolic murmur over the left sternal border. A chest roentgenogram confirmed the presence of dextrocardia, and the ECG was similar to that of twin 1. In contrast to twin 1, there was a 3 cm × 3 cm omphalocele. At 7 hr of age the omphalocele was repaired surgically with an uneventful postoperative course.

Blood type was A+. A karyotype, prepared from peripheral lymphocytes, was 47,XY,+13. The karyotype of twin 1 and twin 2 were identical. In each twin one of the no. 21 chromosomes had a long satellite stalk. In each twin both of the no. 9 chromosomes had a long heterochromatic region. This is a normal variation, but somewhat unusual in both no. 9 chromosomes in a single karyotype.

As in the case of twin 1, after being informed of the cytogenetic diagnosis, the parents refused further diagnostic procedures. The infant died suddenly at the age of 41 days. Consent for autopsy, as in the case of twin 1, was refused.

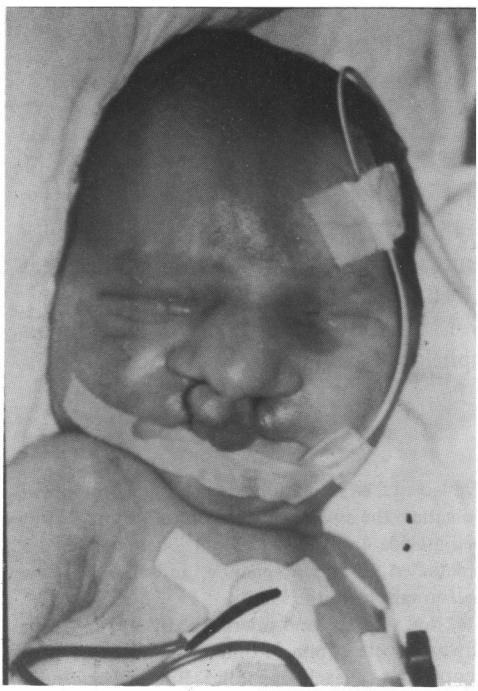

**Figure 1.** Twin 1 shows cleft lip and cleft palate, bulbous nose, small eyes.

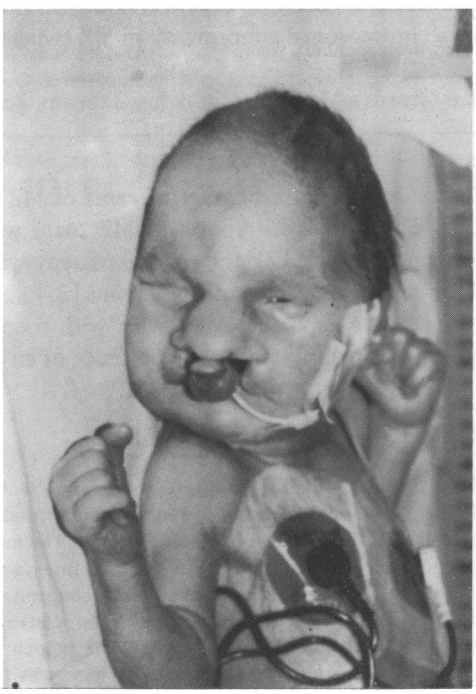

**Figure 2.** Twin 2 shows similar appearance to twin 1.

https://doi.org/10.1017/S0001566000004712 Published online by Cambridge University Press

## DISCUSSION

MZ twins with trisomy 13 appear to be extremely rare. The incidence of trisomy 13 is estimated to be 1/5000 live births. The incidence of MZ twins is 1/250 live births and, in contrast to dizygotic twinning, appears to be independent of race and familial predilection [7]. Therefore, the probability of a live newborn with trisomy 13 being one of a pair of MZ twins is 1/1,250.000. Since there is 95% wastage of fetuses with trisomy 13 [7], the probability of live newborn MZ twins with trisomy 13 is 1/25,000,000 ($4 \times 10^{-8}$).

The incidence of dimorphism in MZ twins is of interest. An epidemiologic survey of congenital malformation in twins was done by Myrianthopoulos [12] as part of a prospective collaborative study of 56,000 pregnant women. There were 1195 twin individuals for whom there was available information. MZ twins had a higher incidence of malformations than singletons or DZ twins for both major and minor abnormalities. The increase in malformations occurred principally in the cardiovascular and alimentary systems with a small increase in central nervous system, musculoskeletal, eye and respiratory malformations. As expected, there was greater concordance in malformations in MZ than in DZ twins.

There was, however, considerable discordance regarding malformations between MZ twins. The discordance rate for 87 pairs of MZ twins with malformations was 72% . Discordance occurred for malformations of the central nervous system in 3 of 4 pairs; for musculoskeletal malformations in 7 of 17 pairs; for eye malformations in 7 of 10 pairs; for upper respiratory tract malformations in all of 4 pairs; for thoracic malformations in 3 of 4 pairs; for cardiovascular malformations in 5 of 6 pairs; for alimentary malformations in 10 of 13 pairs; for liver, bile duct and spleen malformations in 1 of 2 pairs; for genitourinary malformations in 4 of 5 pairs, and for skin malformations in 18 of 21 pairs.

It is very difficult to estimate the incidence of dimorphism in MZ twins with the same chromosomal abnormality because of the rarity of these conditions. The one condition permitting a preliminary survey from published reports on MZ twins is 21 trisomy. We have been able to find reports of 20 such pairs of twins. Of these, 5 pairs were dimorphic for congenital heart disease [5,9,16,20,22]. In some reports insufficient information was present to evaluate dimorhism. There are additional reports of apparently MZ twins with Down syndrome in which authors were hesitant to claim monozygosity because of the presence of dimorphism [11,23].

**Table. Summary of Monozygotic Twins with Chromosome Abnormalities Other than Down Syndrome with Dimorphism**

| Reference | Chromosomal anomaly | Age | Sex | Proof of zygosity | Discordance |
|---|---|---|---|---|---|
| Hata et al [3] | ring 18 mosaicism | 4 yr | M | blood groups | One normal twin; other twin had hypoplastic helices, midface hypoplasia, flat nasal bridge, muscle hypotonia, short stature, mental retardation and retarded growth |
| Reyes et al [17] | trisomy 8 mosaicism | 2 yr | M | placenta, dermatoglyphics | One stillborn twin/dimorphism for renal abnormalities |
| Hustinx et al [4] | 48,XXY, +21 | 7 mo | M | blood | Different cardiac anomalies |

https://doi.org/10.1017/S0001566000004712 Published online by Cambridge University Press

It is possible, moreover, that an abnormal karyotype in each of MZ twins may predispose to discordant anomalies. Preliminary evidence of this phenomenon is shown in the summary of MZ twins with chromosomal abnormalities other than Down syndrome with dimorphism (Table). Our case of MZ twins with trisomy 13 and dimorphism for omphalocele constitutes further evidence. Analysis of additional reports will be required to test this hypothesis.

## REFERENCES


1. Allen G, Baroff GS (1955): Mongoloid twins and their siblings. Acta Genet 5: 294-326.
2. Bencini MA (1952): Contributo clinico allo studio del mongolismo nei gemelli: presentazione di una coppia di gemelle monozigotiche. Acta Genet Med Gemellol I: 29-39.
3. Hata A, Suzuki Y, Matsui I, Kuroki Y (1982): Ring 18 mosaicism in identical twins. Hum Genet 62:364-367.
4. Hustinx TWJ, Eberle P, Geertz SJ, Brink JT, Woltring LM (1961): Mongoloid twins with 48 chromosomes ($AA+A_{21}XXY$). Ann Human Genet 25:111-115.
5. Jervis GA (1943): Mongolism in twins. Am J Ment Defic 47: 364-369.
6. Keay AJ (1958): The significance of twins in mongolism in the light of new evidence. J Ment Defic Res 2:1-7.
7. Kelly TE (1980): Clinical genetics and genetic counseling. Yearbook Med Pub, Chicago.
8. Liberfarb RM, Atkins L, Holmes LB (1978): Down syndrome in two of three triplets. Clin Genet 14:261-264.
9. Lipovetzkaya NG (1964): A case of Down's disease in both uniovular twins. Pediatriia 43:102-103.
10. McDonald AD (1964): Mongolism in twins. J Med Genet 1:39-41.
11. Mikkelsen M, Melchior JC (1962): Mongoloid twins with trisomy of chromosome No. 21. Acta Genet (Base 1) 12:164-171.
12. Myrianthopoulos NC (1975): Congenital malformations in twins, epidemiological survey. New York, Stratton Intercontinental Med. Book Corp.
13. Nance WE, Uchida I (1964): Turner's syndrome, twinning and an unusual variant of glucose-6-phosphate dehydrogenese. Am J Human Genet 16:380-392.
14. Oster J (1953): Mongolism. A clinicogenealogical investigation comprising 526 mongols living in Seeland and neighbouring islands in Denmark. Copenhagen, Danish Science Press.
15. Papp Z (1975): Monozygotic twins with concordance for Down's syndrome and congenital heart disease. Acta Genet Med Gemellol 24:41-46.
16. Rasore-Quartino A (1967): Gemelli MZ con mongolismo concordante. Acta Genet Med Gemellol 16:395-401.
17. Reyes PG, Hsu LYF, Strauss L, Rose J, Hirschhorn K (1978): Trisomy 8 mosaicism syndrome. Report of monozygotic twins. Clin Genet 14:90-97.
18. Shapiro LR, Farnsworth PG (1972): Down's syndrome in twins. Clin Genet 3:364-370.
19. Shiono H, Kasahara S (1971): Genet studies on twins of Down's syndrome with 47-chromosomes. Tohoku J Exp Med 105:309-316.
20. Telfer MA, Baker D, Bergman M (1972): Twins, probably monozygotic, displaying Down's syndrome, physical and functional mirror-imaging, and discordance for congenital heart disease. Am J Ment Defic 76:391-396.
21. Wolff E de, Scharer K, Lejeune J (1962): Contribution a l'étude des jumeaux mongoliens. Un cas de monozygotisme hétérocaryote Helv Paediat Acta 17:301-328.
22. Young RJ (1954): Mongolism in both of monozygotic twins. Arch Dis Child 29:55-59.
23. Zellweger H (1968): Familial aggregates of the 21-trisomy syndrome. Ann NY Acad Sci 155:784-792.


**Correspondence:** Professor H.T. Loevy, College of Dentistry, University of Illinois, 801 South Paulina, Chicago, IL 60612, USA.