47,XX,+13/46,XX MOSAICISM: A CASE REPORT

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We described a D_1 trisomy syndrome patient who had a normal 46,XX cell line and 15.4% of lymphocytes with a 47,XX,+D chromosome constitution as an occasion to review normal trisomy 13 mosaicism.

INTRODUCTION

In man, trisomy 13 leads to a characteristic multiple congenital anomaly/mental retardation (MCA/MR) syndrome which includes postnatal growth failure, usually profound MR and microcephaly at times associated with other overt manifestations of the alobar holoprosencephaly developmental field defect or complex (DFC) such as cebocephaly, midline cleft of lip and palate, etc., frequent eye involvement such as iris coloboma, microphthalmia, rarely anophthalmia OU, postaxial polydactyly, a characteristic complex of minor facial, auricular and dermatoglyphic anomalies, and multiple visceral anomalies affecting the heart, diaphragm, gastrointestinal and genitourinary tract (Patau et al. 1960, Smith et al. 1963, Smith 1964, Warkany et al. 1966, Taylor 1968).

This paper reports 47,XX,+13/46,XX mosaicism in a child with typical features of the 13 (D_1) trisomy syndrome, but who had a normal chromosome constitution in the majority of her lymphocytes.

CLINICAL REPORT

This 3 month-old white female infant was born in January 1967 at term by normal delivery weighing 2400 g; her birth length and occipito-frontal circumference (OFC) are unknown. Parents were normal, not consanguineous, and 23 years old at the time of the child's delivery. The mother was not exposed to unusual amounts of radiation before or after conception of this child. The patient was the youngest of 3 siblings, the other 2 being in good health. At birth the infant did not cry spontaneously, had marked cyanosis and developed respiratory distress, but responded well to resuscitation. Suckling was poor and psychomotor development grossly deficient. At the time of death, she was unable to hold up her head, roll over or sit up. Frequent convulsions responded inadequately to treatment with antiepileptic drugs. She had multiple upper respiratory infections and at least three episodes of acute dehydration, the last being followed by bronchial pneumonia and death. Growth was severely impaired: at 9 months she weighed 4500 g and was 56 cm long. When first examined at 3 months she weighed 3500 g and was 53 cm long. Intermittent periods of apparent respiratory distress were followed by prolonged apneic apells, cyanosis and convulsions. There was an apical systolic murmur presumed to indicate congenital heart disease, a liver edge palpable 1 cm below the right costal margin, profound psychomotor retardation and no response to sound interpreted as severe deafness. There was severe microcephaly (OFC = 29 cm) with prominence of sutures (especially of the coronal metopic sutures) and a minute anterior fontanel, all indicative of early, secondary synostosis due to primary

severe microencephaly. The facial manifestations of alobar holoprosencephaly included: severe hypoplasia of frontal region of skull and of supraorbital ridges, hypotelorium with slight mongoloid slant of palpebral fissures and apparent prominence of globes, absence of nasal bones, nasal septum and premaxilla, a wide midline cleft of upper lip with a narrow bar of tissue delimiting a single nostril from the oral cavity, wide midline cleft of palate. There were minor malformations of the helix present bilaterally; flexion contractures of the 8 lateral fingers which, in each hand, grasped a permanently flexed cortical thumb; umbilical hernia, diastasis recti, generalized muscle hypertony with De Lange's sign (scissoring of the legs) and severe hyperreflexia interpreted as severe congenital spastic tetraplegia. These neurologic manifestations, together with intermittent opisthotonus, apneic spells, convulsions, total lack of psychomotor development and a defect in thermoregulation (with intermittent hypothermia) all confirmed the clinical impression of alobar holoprosencephaly. There was no iris or lens coloboma, no corneal clouding, polydactyly, excessive capillary hemangiomata, external genital abnormalities, jaundice, hyperconvex nails, or a simian crease on either palm. Dermatoglyphics were difficult to study. The right palm showed a t with atd angle of 58°. The left palmar axial triradius could not be discerned. Fingers 1-5 showed respectively U, W, U, W, U on both hands. In the right hallucal area the patterns could not be safely analysed.

Results of routine laboratory studies were normal. Hemoglobin studies at 7 months by starch gel electrophoresis and quantitative determination (Betke et al. 1959, Lehmann and Huntsman 1974) showed 11.2% fetal hemoglobin, 28% hemoglobin A_2 and 86% hemoglobin A_1.

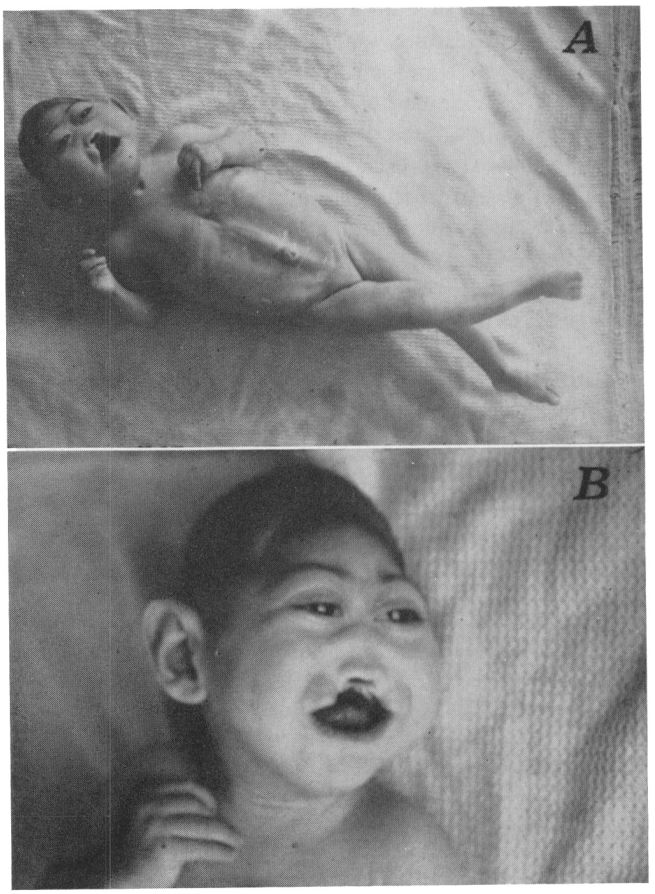

Figure. Propositus, 7 months old.
A: body appearance;
B: close-up of facial appearance.

https://doi.org/10.1017/S1120962300791542 Published online by Cambridge University Press

Roentgenograms of the skull confirmed the clinical impressions of microcephaly and of premature, secondary fusion of sutures. The sella turcica was normal. The rest of the skeleton was osteoporotic with signs of rickets (diphenylhydantoin effect). Bone age was roughly compatible with chronological age. Chest films suggested enlargement of the right ventricle and right atrium, increased pulmonary vascularity, and eventration of the diaphragm. The ECG showed biventricular overload with predominance of the right chambers. Several sleep EEG's showed sharp waves with diffuse bilateral projection especially to the left side. Autopsy was not permitted by the parents.

Chromosome studies used a modified Moorhead technique (Moorhead et al. 1960). Technical difficulties allowed analysis of only 26 mitoses of which 22 had an normal female karyotype; 4 of them (15.4%) had a 47,XX,+D constitution. In view of the characteristic clinical picture the extra D chromosome can be presumed to be an extra 13.

DISCUSSION

The diagnosis of the D_1 trisomy syndrome was easily made on the basis of the clinical phenotype, in spite of the (expected) absence of some manifestations of the syndrome which could either represent normal variability or the effect of mosaicism with a normal cell line (Gerald 1969, Taylor et al. 1970). Mosaic D_1 trisomy cases are listed in Table 1 where their manifestations are compared to those of infants with apparent nonmosaic 13 trisomy. The table shows a greater phenotypic variability of the mosaic cases than is observed in nonmosaic 13 trisomy cases. These data are biased since none of these cases were ascertained in a prospective newborn chromosome study which is the only way to obtain an unbiased ascertainment of *all* D_1 trisomy syndrome cases surviving to birth. Many of the cases in Table 1 were presumably ascertained because of an atypical phenotype.

Table 1. *Sex, birth weight, parental age and chromosomic patterns in regular trisomics and mosaic subjects*

Author [a]	Sex	Birth weight (kg)	Maternal age (at birth)	Paternal age (at birth)	Chromosome analysis
1	35 F, 29M [b]	2.609 ± 86.7 [b]	31.6 ± 0.8 [b]	31.9 ± 1.0 [b]	[c]
2	M	3.033	38	42	46,XY/47,XY,D+
3	F	—	23	23	46,XX,Dp-/46,XX,Di
4 (case 9)	M	4.133	28	28	46,XY/47,XY,D+
5	F	—	34	30	46,XX/47,XX,D+
6 (case 1)	F	3.670	22	22	46,XX/47,XX,D+
6 (case 2)	M	4.024	29	33	46,XY/47,XY,D+
7	F	2.494	31	30	46,XX/47,XX,D+
8	F	3.260	22	23	46,XX/46,XX,C-t(C_qD_q)+
9 (case 2)	M	2.720	18	16	46,XY/47,XY,D+
10	M	3.850	22	22	46,XY/46,XY,E-t(18_qD_q)+
11 (case 1)	F	—	—	—	46,XX/47,XX,D+
11 (case 2)	F	—	—	—	46,XX/47,XX,D+
11 (case 3)	F	—	40	—	46,XX/47,XX,D+
12 (case 3)	F	2.830	26	29	46,XX/46,XX,D_qi
13	M	—	35	—	46,XY/47,XY,D+
14	F	2.400	23	23	46,XX/47,XX,D+

[a] 1. Taylor 1968; 2. Warkany et al. 1962; 3. Therman et al. 1963; 4. Taylor and Polani 1964; 5. Forteza et al. 1964; 6. Bain et al. 1965; 7. Stone et al. 1966; 8. Wilson and Melnyk 1967; 9. Engel et al. 1967; 10. Craig and Luzzatti 1967; 11. Green et al. 1968; 12. Taylor et al. 1970; 13. Scouras et al. 1972; 14. Present case.
[b] Data based on 74 cases from literature.
[c] 19 cases 47,D+; 3 cases 46,tD/D; 3 cases with various aberrations; 2 cases with normal chromosomes.

https://doi.org/10.1017/S1120962300791542 Published online by Cambridge University Press

Table 2. *Malformations present in regular trisomic individuals and normal/trisomy mosaics*

Author [a]	% trisomic cells	Microcephaly	Low-set ears	Abnormal ear lobulation	Hare lip	Cleft palate	Ogival palate	Micrognathia	Ocular hypertelorism	Epicanthic folds	Cataracts	Dislocated lenses	Microphthalmia	Iris coloboma	Strabismus	Large, broad nose	Cutis laxa	Polydactyly	Flexion deformity (fingers)	Hyperconvex nails	Retroflexible thumbs	Equinovarus feet
1 [b]	—	64	92	80	58	69	0	84	92	56	0	0	76	33	6.7	0	0	76	68	37	25	17
2	70	+	+	+	—	—	+	+	—	—	—	—	—	—	+	—	0	—	+	0	—	0
3	55	+	—	—	+	+	+	—	—	+	—	—	—	+	—	—	—	—	—	+	—	—
4	< 2	+	+	+	—	—	—	—	—	—	—	—	—	—	—	—	0	—	—	0	—	—
5	36	0	+	—	—	+	+	+	—	—	—	—	—	—	—	0	0	—	—	—	—	—
6	22	—	—	+	—	+	+	+	—	—	—	—	—	—	—	0	0	—	+	—	—	—
6	40	—	+	0	—	—	—	—	—	—	—	—	—	—	—	0	0	—	+	—	—	—
7	15	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	0	0	—
8	0	+	—	—	+	+	+	—	—	+	+	—	+	+	—	+	0	—	+	—	—	—
9	<90	—	+	0	—	—	+	—	—	+	+	+	—	+	—	+	0	—	+	0	0	—
10	70	+	+	0	—	—	+	0	—	—	—	—	+	+	—	0	0	+	—	—	—	—
11	42	0	0	0	—	—	+	+	+	+	—	—	—	—	0	—	0	0	—	0	0	—
11	28	—	+	0	—	—	+	+	—	—	—	—	—	—	—	+	+	—	—	0	0	—
11	30	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
12	<50	+	+	0	+	+	+	+	—	—	—	—	—	—	—	0	0	+	—	0	+	—
13	32	+	+	—	—	—	+	—	—	+	+	—	—	—	+	—	—	+	—	—	—	—
14	15	+	+	+	+	+	+	+	—	—	—	—	+	—	—	+	—	—	+	+	0	—

+ feature present; — feature absent; 0 not stated.

[a] Same as Table 1.

[b] Percent of feature among D_1 trisomics.

https://doi.org/10.1017/S1120962300791542 Published online by Cambridge University Press

Table 2. *Contd.*

Calcaneovalgus feet	Capillary hemangiomata	Diastasis recti	Inguinal hernia	Umbilical hernia	Hypogenitalism	Brain malformation	Congenital heart malformation	Renal malformation	Hypertonia	Hypotonia	Impaired hearing	Seizures	Psychomotor retardation	Other malformations
8.7	72	0	40	00	93	70	76	48	26	48	25	50	100	
0	—	—	—	—	+	0	—	+	—	—	—	—	+	Cranial, facial and lower member asymmetry; spina bifida; absent patellae; camptodactyly; alternating esotropia.
—	+	+	—	+	—	0	—	0	—	—	—	—	+	
—	—	—	—	—	0	0	0	0	—	—	+	—	+	
—	—	—	—	—	0	+	—	0	0	0	+	+	+	Café-au-lait spots; malpositioned teeth.
—	—	—	—	—	+	+	+	—	—	+	—	0	+	
—	—	—	—	—	—	—	+	—	0	0	—	0	+	Large head; coarse facies; deep winkling of brow.
—	—	0	—	—	—	0	—	0	—	—	—	+	±	Asymmetry of maxilla; deviation of sacrum.
—	+	0	—	—	—	+	—	+	+	—	+	0	+	High forehead.
—	—	0	—	—	+	0	—	0	+	—	+	+	+	Sloping forehead.
—	—	—	—	—	+	0	+	0	—	—	—	+	+	Dislocation of hips; sloping forehead; pyloric stenosis.
—	—	—	—	—	—	0	+	0	—	—	—	—	—	Acne of head and neck; protrunding sternum; kyphosis; long tapering fingers.
—	—	—	—	—	—	0	+	0	—	—	—	—	±	Hydrocephalus; lumbar lordosis; widely spaced nipples; short hands and feet.
—	—	—	—	—	—	—	—	—	—	—	—	—	—	Schizophrenia.
+	—	—	—	—	—	+	+	+	0	0	0	0	+	
—	—	—	—	—	+	+	+	0	—	+	—	—	+	Bifid teeth; nystagmus.
+	—	—	—	+	0	0	+	0	+	—	+	+	+	

https://doi.org/10.1017/S1120962300791542 Published online by Cambridge University Press

Since no autopsy was done on our patient, only inferences are possible concerning the possible presence or absence of some internal congenital malformations commonly seen in the D_1 trisomy syndrome. On the basis of the cranio-facial and the neurological manifestations a diagnosis of alobar holoprosencephaly seems virtually certain, though unusual transillumination of the skull/brain would have helped with that diagnosis (DeMeyer and Zeman 1963, DeMeyer et al. 1963, DeMeyer et al. 1964). It must be stressed this DFC is an etiologically nonspecific anomaly and may occur as Mendelian traits (both autosomal recessive and autosomal dominant forms of alobar holoprosencephaly are known (Dellaire et al. 1971) and component manifestations of other chromosomal MCA/MR syndrome such as the 18p and 13q syndromes (Uchida et al. 1965, Opitz et al. 1969). Brain malformations are frequently found in the D_1 trisomy syndrome (Taylor 1968).

Data from the cardiological examinations suggest the presence of acyanotic congenital heart disease, most likely a patent ductus arteriosus which are the two most common types of congenital heart defect seen in the D_1 trisomy syndrome (Warkany et al. 1966, Taylor 1968).

No inference gastrointestinal and genitourinary tracts are possible since contrast studies were not done. The fetal hemoglobin levels are elevated (Jonxis and Visser 1956).

Similar changes have been noted before in the D_1 trisomy syndrome (Huehns et al. 1964) and probably represent dyssynchronous maturation (Lee et al. 1966).

Distal displacement of triradius t when compared with a normal Brasilian control group (Toledo et al. 1969) occurs commonly in the D_1 trisomy syndrome. Typical hallucal pattern of this entity (Uchida et al. 1962) could not be discerned in the present case.

The frequency at birth of the D_1 trisomy syndrome is estimated from 1 in 7000 (Taylor 1968) to 1 in 14,500 (Conen and Erkman 1966). Combining data from different estimates, the mean frequency at birth is about 1 in 10,000 (Berger 1973, Jacobs et al. 1974). About 5% of them are thought to be mosaics with a normal cell line (Magenis et al. 1968). Some 50 mosaic cases have been reported (Magenis et al. 1968, Uchida et al. 1962, Huehns et al. 1964) including the 15 cases summarized in Table 1 (Warkany et al. 1962, Therman et al. 1963, Stone et al. 1966, Craig and Luzzatti 1967, Engel et al. 1967, Scouras et al. 1972). We ruled out from this compilation cases with similar mosaicism but features suggesting non-involvment of a 13 chromosome. Therefore, we excluded patients with otosclerosis and 4 normal relatives (Tato et al. 1963); 4 cases of congenital analgesia from 3 different families (Becak et al. 1964); 3 propositi with congenital heart disease (Domingo-Sanz 1966) and a case with gynecomastia, pectus excavatum and lumbar kyphosis (Berger et al. 1973). We also excluded cases with apparent D_1 trisomy syndrome manifestations but with suggested normal chromosomes (DeMeyer et al. 1963, Marshall et al. 1964, Taylor et al. 1970, Dellaire et al. 1971) or with unrelated chromosome abnormalities (Uchida et al. 1965). Apparent D_1 trisomy syndrome patients with normal chromosome may have the Meckel syndrome (Opitz and Howe 1969).

The cases in Tables 1 and 2 include two daughters with minor anomalies born to a phenotypically normal woman; all three had a mosaic 47,XX,+D/46,XX constitution and trisomy 13 was confirmed by autoradiographic studies (Green et al. 1968). The cause of such familial mosaicism is unknown and a " genetic tendency to nondisjunction " is a frequently suggested hypothesis to explain such occurrences.

Since the patient died in 1968 and the available slides did not permit further analysis, banding techniques were not performed. In spite of this, all the evidences are favoring the hypothesis that the extra D chromosome was from group 13. Furthermore, trisomy 14 and 15 are mainly

found in miscarriages (Conen and Erkman 1966, Jacobs et al. 1974) and their clinical phenotypes do not overlap with the one here reported.

The proportion of the two cell lines varies from patient to patient and does not seem to bear a relationship to clinical severity. Thus, our patient with a rather low frequency of trisomic cells is one of the most severely affected of mosaic cases. Different proportions of the 2 cell lines in different tissues of the same patient have been reported (Forteza et al. 1964, Bain et al. 1965, Wilson and Melnyk 1967) and may reflect selection during growth and development. Although data on mosaic subjects are based on a small number of individuals, we find that mean parental age does not differ significantly from that of the normal population or from that of propositi with a D_1 translocation (Taylor 1968, Taylor et al. 1970, Jacobs et al. 1974). This suggests that most 13 trisomy mosaics arise by mitotic nondisjunction in normal zygotes. In the present case both parents were also young (i.e., 23 years). Except for the familial mosaic cases no genealogical preconceptional or epidemiological causes of 13 trisomy/normal mosaicism are known.

Mosaic D_1 trisomies generally live longer than most regular trisomic subjects (Taylor et al. 1970, Magenis et al. 1968). Only 12.2% of D_1 trisomy infants reaches the age of 1 year, whereas over 1/3 the mosaics is still alive at that age. Our patient died at 13 months.

Acknowledgements

It is a pleasure to record our thanks to Dr. John M. Opitz for his critical review and editing of the manuscript. This paper was partially supported by GNPQ (Brasil), SIP - 04/11.

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RIASSUNTO

Mosaicismo 47,XX,+13/46,XX: Un Caso Clinico

Viene descritto un paziente affetto da sindrome da trisomia D_1, con una linea cellulare normale 46,XX ed il 15,4% dei linfociti con costituzione cromosomica 47,XX,+D. Con l'occasione, viene effettuata una rassegna del mosaicismo da trisomia 13.

RÉSUMÉ

Mosaïcisme 47,XX,+13/46,XX: Cas Clinique

Un patient atteint de syndrome de trisomie D_1 (15,4% de lymphocytes à constitution 47,XX,+D) est décrit et une revue de la condition est effectuée.

ZUSAMMENFASSUNG

47,XX,+13/46,XX-Mosaik: ein klinischer Fall

Beschreibung eines Patienten mit D_1-Trisomie-Syndrom, bei dem eine normale Linie aus 46,XX-Zellen besteht, während 15,4% der Lymphozyten eine Chromosomen-Bildung von 47,XX,+D aufweisen. Bei der Gelegeneheit wird eine Übersicht über das Trisomie-13-Mosaik gegeben.

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https://doi.org/10.1017/S1120962300791542 Published online by Cambridge University Press