Letter to Editor

The second case with 47, XY, + 8 [38] / 45, X0 [12] karyotype

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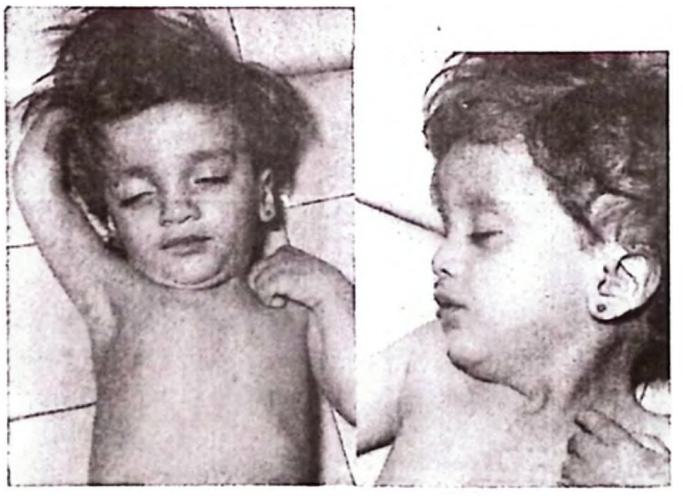
Trisomies are the cause of more than 50% of all spontaneous abortions[1]. In the literature, trisomy 8 is more frequently reported as mosaic cases with normal karyotype[2,3]. Features of mosaic trisomy 8 show great variability such as mental retardation in 90%, malformed pinnae, and contractures of the fingers and toes in 70%, vertebral anomalies in 55%, (which show progression), as the individual gets older, hypertelorism and strabismus in 50%, broad and upturned nose with anteverted nostrils and high palate in 60%, micrognathia, and everted lower lip in 40%, deep furrows on palms and soles in 75%, urogenital anomalies in 40%, and congenital cardiac anomalies in 25%[4,5].

Turner syndrome is characterized by short stature, primary amenorrhea, webbed necked, cubitus valgus in postpubertal females and sexual infantilism. Mental status is expected to be normal. Other common findings are epicanthal folds, ptosis of upper eyelids, prominent ears and micrognathia, and low-set hair-line in 75%; wide-spaced, hypoplastic nipples in 60%; small for gestational age in 50%; excess skin on the nape and peripheral lymphedema in infancy in 40%; hypoplastic, hyperconvex and deep-set toenails in 75%; increased numbers of cutaneous nevi in 60%; aortic stenosis and idiopathic hypertension in 25%; renal anomalies in 40%; and deafness in 50%[4].

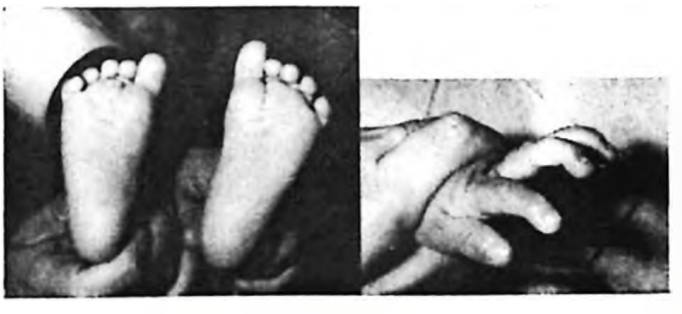
Turner syndrome and mosaic trisomy 8 have been reported in the literature by DeBrasi et al.[6] in 1995; we present the second case with those cytogenetic anomalies.

A twenty-three-month-old female case was admitted to the hospital because of delayed motor and mental development. She was born at term to non-consanguineous parents after a normal vaginal delivery. Her birth weight and height were not recorded. She had meningitis in the first year of life and chronic otitis.

On admission, she was 7,400 g (< 3rd centile), and 72 cm (< 3rd centile). Her head circumference was 46 cm (2-50 centile). She had protruding forehead, long face, hypertelorism, broad, upturned, and bulbous nose, long upper lip, thick and everted lower lip, micrognathia, low-set and deformed ears, high palate, short and broad neck, wide-spaced nipples, arachnodactyly, camptodactyly, deep furrows on palms and soles, and severe motor and mental retardation (Fig. 1a and 1b). Her blood analysis showed hypochromia and microcytosis. Her urine analysis, liver and renal functions were normal.



(a)



(b)

Fig. 1. a) Features of case at 23 months of age. b) Fingers and soles of case.

Her ECG, telecardiography and echocardiography were normal. Abdominal ultrasound (US) examination revealed ovarian agenesis. X-ray examination showed broad ribs and bone age delay. Her karyotype was 47, XY,+8 [38] / 45,X0 [12] (Fig. 2a and 2b). Parents' karyotypes were normal.

DeBrasi et al.[6] reported two cases with mosaic trisomy 8 syndrome and sexual chromosome aneuploidies. Karyotype of the first case was 45,X[59.2%] / 46,X,+8[1.2%] / 47, XX, + 8 [39.6%] and of the second case was 47, XX, + 8 [61.7%] / 47, XXY [38.3%]. They studied molecular analysis to explain the mechanism of those complex karyotypes and found postzygotic mitotic errors.[6] Karadima et al.[7] showed postzygotic mitotic errors by molecular analysis in cases with autosomal trisomies contrary to the common view that maternal meiotic errors cause autosomal trisomies.

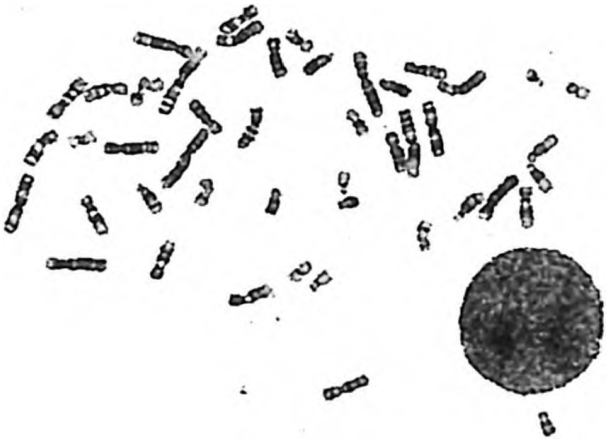


Fig. 2a. Trisomy 8 cell line of case.

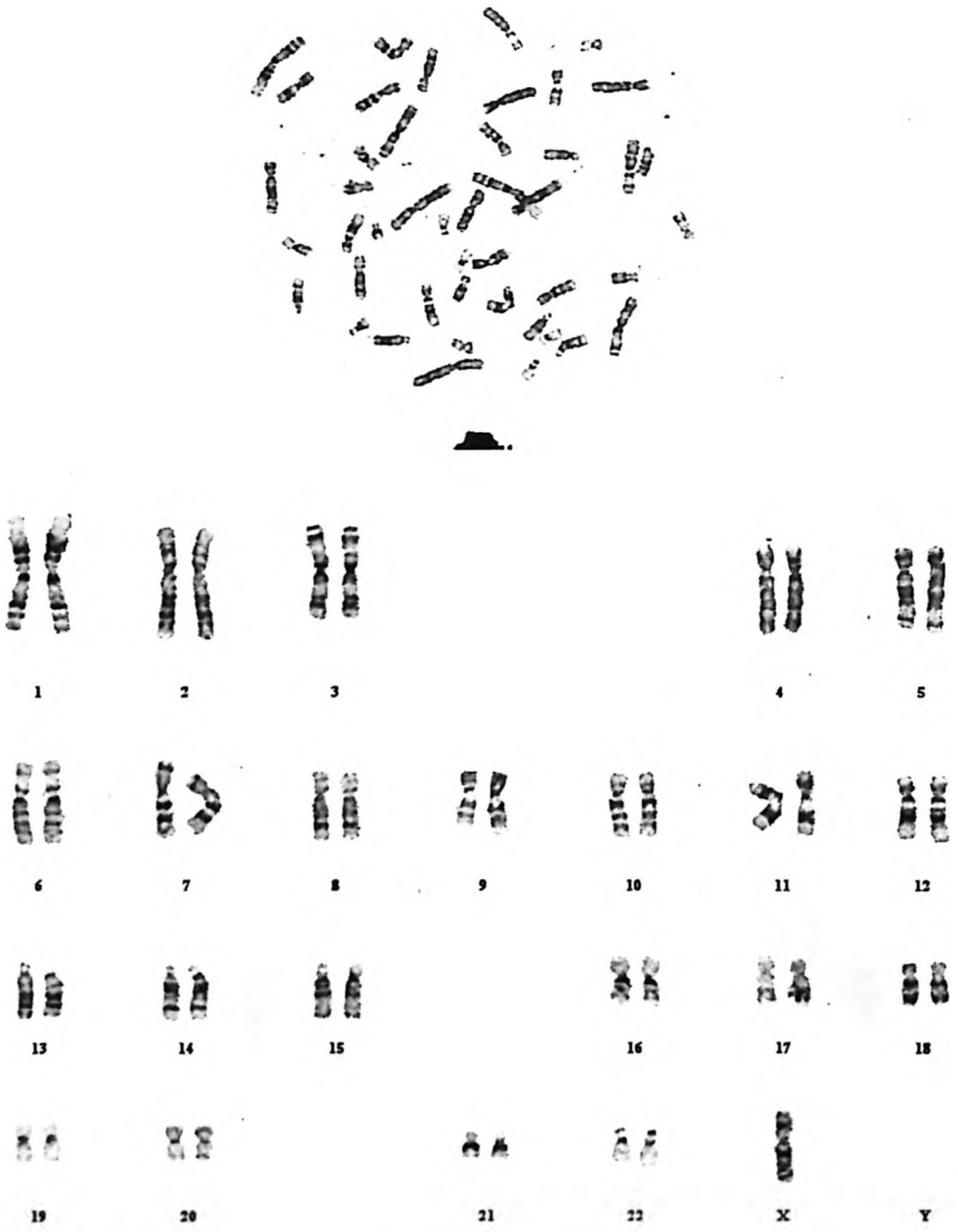


Fig. 2b. 45, XO cell line of case.

The most common findings of mosaic trisomy 8 such as protruding forehead, long face, hypertelorism, anteverted nostrils and broad nose, long upper lip, thick and everted lower lip, micrognathia, dysplastic and low-set ears, high palate, arachnodactyly and camptodactyly, and deep furrows on palms and soles were detected in our case as well. Short, broad neck, wide-spaced nipples and ovarian agenesis detected by US can be considered as the features of Turner syndrome. No cardiac anomaly was detected in our case; however, it can be seen in both trisomy 8 and Turner syndrome cases.

Cardiac and renal anomalies are the major factors that affect the prognosis in both trisomy 8 and Turner syndrome. The patient presented here is the second case in the literature with trisomy 8 and Turner mosaicism showing the most common features of both cytogenetic abnormalities.

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