typic expression in families with identical X chromosome abnormalities and non-random inactivation are the result of the differences in the genetic content of the normal, active X.⁷ The mental retardation or dysmorphic features or both in the proband could, therefore, be the result of the expression of genes on the normal X which is active in all the cells.

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Martsolf's syndrome in a non-Jewish boy

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SUMMARY Martsolf's syndrome has been described in Jewish people. We describe a patient of non-Jewish ancestry who has minor differences from other patients. The possible pattern of inheritance is discussed.

In 1978, Martsolf *et al*¹ described two brothers, born to consanguineous Jewish parents, with severe mental retardation, cataracts, short stature, and primary hypogonadism. Another Jewish family with the same syndrome has recently been described.² We describe here a non-Jewish boy, to clarify the clinical picture of this syndrome further and to determine its mode of inheritance which is still uncertain.

Case report

The proband was a 13 month old male, born to a G1 P1 A0 20 year old woman after an uncomplicated term pregnancy. There was no exposure to any known teratogenic agents during pregnancy. Delivery was uncomplicated and spontaneous. His birth weight was 2100 g. The parents, of non-Jewish origin, were healthy and non-consanguineous.

There was no family history of other congenital malformations. At three months of age, the patient was admitted to hospital because of bronchopneumonia and retardation of growth. On this

occasion bilateral cataracts were noted and at the age of seven months he underwent a surgical operation for these. At 13 months of age, he was admitted to our hospital because of growth and psychomotor retardation. On admission, physical

FIG 1 The proband at 13 months of age.

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FIG 2 Genitalia of the proband at 13 months of age.

examination showed a boy with dysmorphic features (fig 1). His weight was 7000 g (<3rd centile), length 74.4 cm (10th centile), and head circumference 42.5 cm (<3rd centile). He had brachycephaly, flat occiput, low nasal bridge, mild micrognathia, low posterior hairline, normal philtrum, normal tongue, absence of hypotelorism, and a high arched palate. Pectus excavatum with normal nipples and mild lumbar lordosis were present as well as lax finger joints. Dermatoglyphic analysis showed absence of simian lines on the palms which were otherwise normal and six whorls and four ulnar loops on the fingertips; the toenails were normal. Bilateral

cryptorchidism was present (fig 2), but the penis was normal (stretched penile length 5 cm). Neurological evaluation showed psychomotor development corresponding to six months of age. X ray and cardiovascular studies were normal. TORCH and urinary amino acid, mucopolysaccharide, and oligosaccharide patterns were normal. Basal values of FSH (2.7 mUI/ml), LH (5.7 mUI/ml), and testosterone (10 ng/dl) were normal for age. Leydig cell reserve after HCG stimulation test (2000 UI $\times$ three days)³ was normal (testosterone 200 ng/dl). Chromosome analysis showed a normal male karyotype.

Discussion

Martsolf's syndrome with its rather non-specific features is a very difficult syndrome to diagnose. However, the presence of severe mental retardation, cataracts, short stature, microcephaly and bilateral cryptorchidism suggest that our patient has the syndrome, despite some minor differences (table). In our young patient the principal clinical signs of the syndrome are already present and his dermatoglyphic pattern is similar to those of other patients.¹ However, he differs from some of the patients described previously^{4,5} in whom other clinical signs were present besides the cataracts, mental retardation, and hypogonadism.

Our patient, although having bilateral cryptorchidism, has a normal reserve of Leydig cells so far; follow up will be necessary to show primary

TABLE Summary of clinical findings.

Clinical findings	Martsolf et al ¹		Sanchez et al ²		Our patient
	Patient BS	Patient SS	Case 1	Case 2	
Severe mental retardation	+	+	+	?	+
Height 3rd centile	+	+	+	-	-
Weight 3rd centile	+	+	+	-	+
Head circumference 3rd centile	+	+	+	+	+
Brachycephaly	+	+	+	+	+
Aged appearance	+	+	+	?	-
Cataracts	+	+	+	+	+
Hypotelorism	+	-	-	-	-
Low nasal bridge	?	+	-	-	+
Maxillary retrusion	+	+	+	+	+
Short philtrum	+	-	+	-	-
Malaligned teeth	+	+	+	?	?
Furrowed tongue	+	+	-	?	-
Sparse facial hair	+	+	?	?	?
Low posterior hairline	+	-	+	?	+
Flat broad sternum	+	+	+	+	-
Prominent nipples	+	+	-	-	-
Lax finger joints	+	+	+	+	+
Short palms	+	+	+	+	-
Lumbar lordosis	+	+	+	+	+
Abnormal toenails	+	+	-	-	-
Talipes valgus	+	+	+	+	+
Hypogonadism	+	+	+	+	?
	(primary)	(primary)			

involvement of the gonads. In contrast, the patients of Martsolf *et al*¹ showed clear evidence of primary hypogonadism. Possible hypogonadotrophic hypogonadism cannot be ruled out as a cause of hypogonadism in the patients of Sanchez *et al*.² Finally, our case, the only patient of non-Jewish ancestry reported so far, points to the likelihood that this syndrome does not affect only Jewish people. The fact that our patient is also male suggests two possible patterns of inheritance: X linked recessive inheritance or autosomal recessive inheritance with expression limited to males. However, the fact that only males seem to be affected might be a coincidence, since very few cases have been reported. It is evident that further reports are necessary in order to establish the exact pattern of inheritance, to clarify the type of hypogonadism and the clinical picture of this syndrome, and to discover whether this new syndrome affects mainly Jewish people.

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