Familial occurrence of Turner syndrome in two Tunisian families

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Introduction :

Turner syndrome (TS) is a common genetic disorder with an incidence of 1 in 2500 live births due to chromosomal errors resulting in monosomy for the X chromosome with or without mosaicism. Familial TS has been rarely reported. We report two families having TS.

Patients and methods :

We report 6 patients with TS who had been referred to the Endocrinology department and Pediatric department at Hedi Chaker hospital, Sfax, Tunisia. We performed biochemical analysis, imaging and cytogenetic analyses.

Results:

We report two families having TS. In the first, 4 sisters belonging to a consanguineous family (I) were diagnosed at the age of 14, 17, 31 and 43 years, for TS because of the short stature dysmorphic syndrome and delayed puberty. The cytogenetic analyses performed showed different karyotypes 45XO, 45XO/46XX and two had 45X/46XX/47XXX and mother's karyotype analysis revealed no chromosomal abnormality. The second family (II) included monozygotic twins having the same formula 45X/46XX. Their mother karyotype was not analysed. They were diagnosed at the age of 1 year because they suffered from dysmorphic syndrome. They had stature delay and both of them are now under growth hormone treatment.

Table I: Clinical characteristics of our cases according to cytogenetic finding

	Family I				Family II	
	case1	Case 2	case3	Case 4	Case 5	Case 6
Age (years)	57	40	46	48	3,5	3,5
Age at diagnostic	31	14	17	43	1	1
Height cm (SD)	136 (-4 SD)	144 (-3SD)	147 (-2SD)	156 (-1SD)	89 (-1,5)	85 (-1,5)
Dysmorphyc Syndrome	round face micrognathia short neck pigmented noevi short fourth metacarp low hair implantation	short neck pigmented noevi short fourth metacarp low hair implantation	micrognathia short neck short fourth metacarp low hair implantation	cubitus valgus, short neck. low hair implantation	Epicanthus Pterygium colli Short neck low hair implantation	short fourth metacarp Epicanthus Pterygium colli Short neck low hair implantation
TANNER Stage	IV	III	IV	IV	I	I
Karyotype	45X0 (8%)/46XX	45X0 (23%)/46XX /47XXX (3%)	45X0	45X0 (13%)/46XX /47XXX (4%)	45X/46XX	45X/46XX

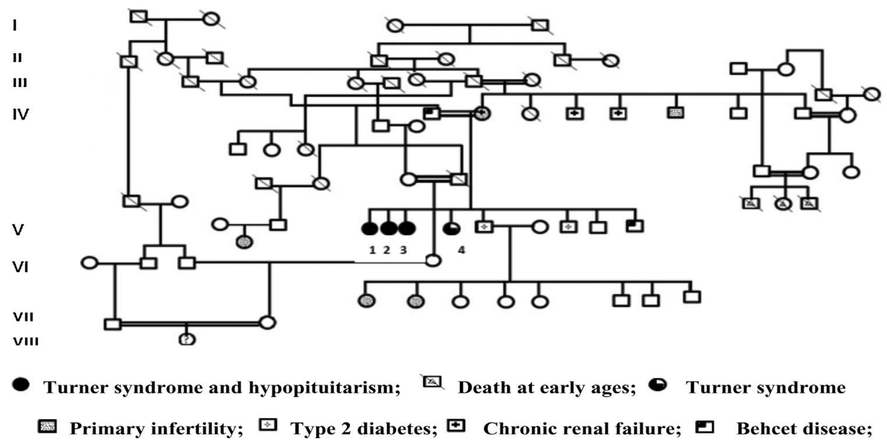

Fig. 1 . Pedigree of the family I

Discussion et conclusion:

The earliest occurrence of familial TS was made by Josso et al. in 1963 in an aunt and her niece with karyotype mosaicism. But the longest number of cases interested seven women with Turner syndrome in a three generations family. Therefore; the current study is the first one reporting of a familial TS in four sisters considering the first family.

Hiromi Muta et al. suggested that some chromosomal abnormalities including mosaicism and genetic defect can be transmitted hereditarily to the descendents in those familial cases and predispose to chromosomal fragility that yields to chromosomal aneuploidy. In our family, mother's karyotype analysis revealed no chromosomal abnormalities. TS in both twin members has been reported eight times, all in monozygotic twins. In TS twin with 45, and 45,X/46,XX, the abnormal chromosome constitution was explained as it has arisen as a result of mitotic nondisjunction in the blastomeres of an originally 46,XX zygote followed by twinning.

Specific maternal karyotype should be taken into account in genetic counselling regarding potential risks for offspring. These cases suggest that a risk of recurrence is possible. And because of the clinical implications, TS families should be studied to exclude familial transmission.

References

(1) Maeda T, Sugano M, Guan JZ, Oyama J, Higuchi Y and al. Familial Turner mosaicism 46XX/45XO with brain calcification. J Neuropsychiatry Clin Neurosci. 2007 Summer;19(3):342-3.

(2) Josso n, De grouchy j, Frezal j, Lamy m, et al. Familial turner's syndrome. Study of 2 families with xo and xx karyotypes. Ann Pediatr (Paris). 1963 Apr 2;10:163-7.