



45,X/46,Xidic(Y) mosaicism:

study of nine patients

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Background: The isodicentric Y (idic Y) is one of the most common aberrations of the Y chromosome. Most patients (pts) are chromosomal mosaics, including 45,X cell line.

Objective: our aim is to describe clinical and molecular features of our 45,X/46,Xidic(Y) cases

Methods: We retrospectively evaluate the clinical description of 9 cases (6 females, 1 male, 2 with ambiguous genitalia) with mosaic karyotype 45,X/46,Xidic(Y) referred to our Centre in the last 23 years.

Patients

Pts	1	2	3	4	5	6	7	8	9
Age of first evaluation	11.8 yrs	1.8 yrs	8 yrs	13.5 yrs	10.6 yrs	16.3 yrs	0.33 yrs	0.16 yrs	27.5 yrs
Sex of rearing	F	F	F	F	F	F	F (AG)	M	M (AG)
Turner's stigmata	+	++	++	+	+	++	++	-	++
Stature	-1.5 SDS	0 SDS	- 5.3 SDS	-3.6 SDS	-2.6 SDS	-1.03 SDS	+1 SDS	0 SDS	-4.1 SDS
Horseshoes kidney	-	+	+	+	-	+	-	+	+
Congenital heart disease	+	+	-	+	+	+	+	-	-
External genitalia	Normal female	Normal female	Normal female	Clitoral hypertrophy	Clitoral hypertrophy	Normal female	Hypospadias Clitoral hypertrophy Cryptorchidism	Hypospadias Cryptorchidism	Hypospadias Cryptorchidism
Internal genitalia	Normal female	Normal female	Normal female	Normal female	Normal female	Normal female	Mullerian remnants, Wolffian structures, Urogenital sinus	Wolffian structures	Mullerian remnants Wolffian structures
Peripheral Karyotype	45X(40%)/ 46,X idic(Y)	45,X(62%)/ 46,X+mar. ish idic(Y)	45,X(79%)/ 46,X+mar idic (Y)	45,X(83,7%)/ 46,X idic(Y)	45,X(97%)/ 46,X idic(Y)	45,X(22%)/ 46,X, psu idic(Y)	45,X(93%)/ 46,X psu idic(Y)	45,X(100)/ 46X,+mar(9).is h idic Y)	45X(56%)/ 46,X psu dic(Y)
Gonad karyotype	L:45X(93%)/ 46,X psu idic (Y) R:45X(98%)/ 46,X psu idic (Y)	NP	NP	NP	NP	NP	L:45X(51%)/ 46,X psu idic (Y) R:45X (79%)/46,X psu idic (Y)	L: Not performed R:45,X(77%)/ 46,X,+mar(12) .ish idic	NP
Gonadal histology	Bilateral streaks	Testis/ streak	Bilateral streaks	NP	Bilateral streaks	NP	Testis/ Streak	dysgenetic testis	Bilateral dysgenetic testes

Four pts (3,4,5,9) with Turner syndrome stigmata, short stature and other malformations showed a predominance of peripheral 45,X cell line. Pt 1 showed the same clinical features but a predominance of peripheral 46,X idic(Y); pt 7 showed complex phenotype despite a predominance of peripheral 45,X. Gonadal histology in cases 3,5,9 correlates with peripheral karyotype. However in pts 1,7 gonadal histology and phenotype correlate more with gonadal than peripheral karyotype.

Conclusion: phenotypic variability and sexual differentiation of these cases are explained by the **degree of mosaicism**, particularly in gonads. To **improve the genotype/phenotype correlation** is useful to analyze more than one tissue and to perform **histological and karyotype studies of the gonads** when it is possible.