SEX CHROMOSOME MOSAICISM IN MALE PATIENTS – OUR EXPERIENCE

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INTRODUCTION: 45X0/46XY mosaicism is rare , with an incidence of 1.7/10,000 pregnancies. It has varied phenotypic spectrum ranging from short stature, ambiguous genitalia, clinical signs of Turner's syndrome in both males and females and normal male/female phenotype.

OBJECTIVES: To describe the clinical characteristics in 6 phenotypical male patients.

METHODS: We retrospectively evaluated the clinical description of 6 patients.

RESULTS: Two patient presented with short stature ,two of them were antenatally diagnosed and two had ambiguous genitalia. The patient characteristics are described below. None of them had renal or cardiac anomalies.

Patients	1	2	3	4	5	6
Antenatal diagnosis	No	No	Yes -previous sibling with Trisomy21	Yes, Nasal glioma	No	No
Age at presentation	12.8yrs	8 yrs	Birth	Birth	Birth ,Preterm,30 wks	Birth
Presentation symptom	Short stature	Short stature			Ambiguous Genitalia	Ambiguous Genitalia
Stature	- 3 SDS	-2 SDS	-1 SDS	0 SDS	-2 SDS	-3 SDS
Turner's stigmata	No	No	No	No	No	No
External Genitalia	Normal male	Normal male	Normal male	Normal male	R testis- Scrotum L testis streak(inguinal) Severe hypospadias with peno-scrotal web	R Testis- Scrotum L testis –Scrotum Hypospadiasis with Chordee Duplication of urethra.
Peripheral Karyotype	45XO/46XY	45XO/46XY	45XO46XY,46X Ring Y chromosome (esoteric)	45XO/46XY	45XO/46XY mosaic	45XO/46XY
Growth hormone Treatment	Yes , 13 yrs.	Yes,9 yrs.	No	No	No	Yes ,4.5 yrs.
Pubertal Induction	Yes	Yes	No	No	No	No

CONCLUSION: In summary, karyotype analysis should be mandatory when investigating short stature in males outside of target centile range particularly those with urogenital anomalies, as is recommended in short stature for girls. These children should be followed up regularly as they tend to respond well to growth hormone treatment but also to monitor for late onset problems such as gonadal tumours and infertility.

REFERENCES:

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