A Clinical and cytogenetic study of patients with Disorders of Sex Development (DSDs) Associated with Congenital Anomalies or Recognizable Syndromes

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Introduction

DSDs represent a **diverse group** of clinical conditions with a very wide phenotypic/genotypic spectrum.
They are considered among the most common birth defects and are frequently associated with **congenital abnormalities.**

Objective

Using advanced cytogenetic techniques like **ACGH in cases of DSD with multiple congenital anomalies for proper diagnosis and counseling**

Patients and methods

62 patients with DSD associated with somatic anomalies. from clinical genetics dept., NRC. underwent:

- Complete **clinical assessment**, Quigley scoring of external genitalia and pubertal staging.
- **Chromosomal analysis** for all patients
- **FISH and ACGH analysis** were performed whenever indicated

Results

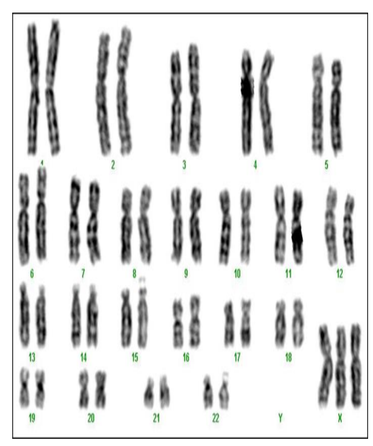
Conventional GTG banding in 49 cases

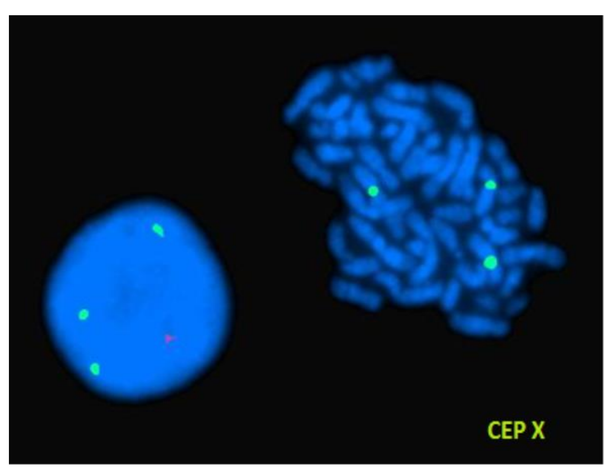
Presentation	Karyotype	No.
Genital anomalies associated with dysmorphism or other congenital anomalies	46,XY	26
	46,XX	15
	49,XXXXY	3
	47,XXY	2
	47,XXX	3

FISH analysis in 13 cases

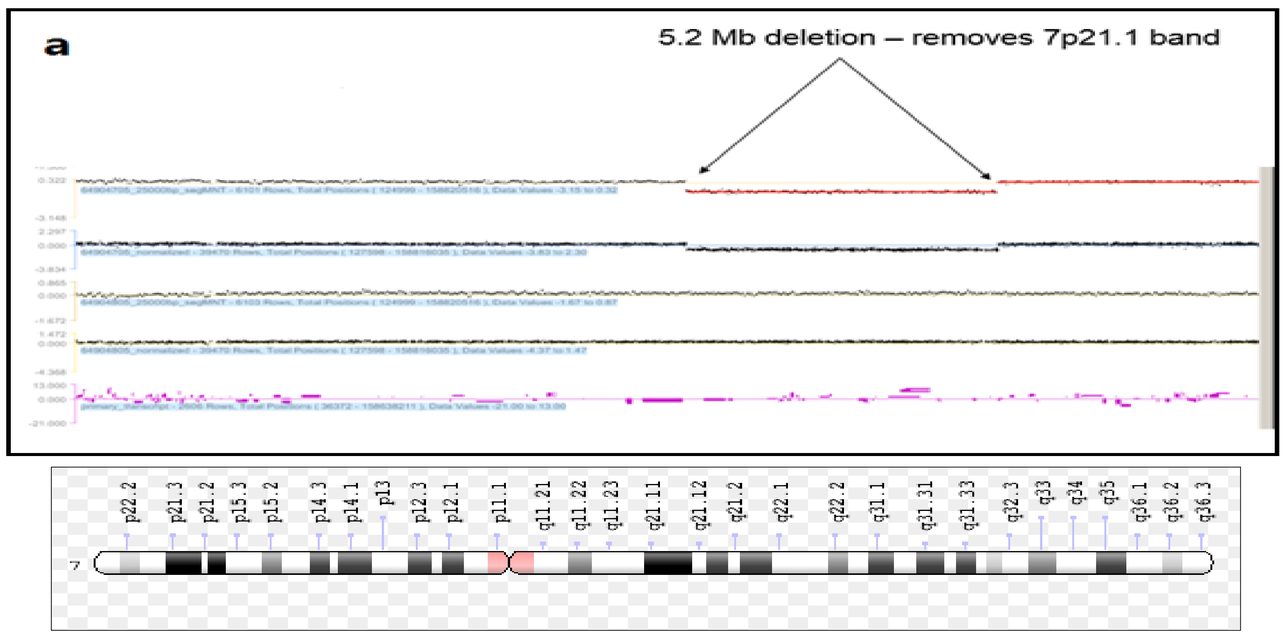
Karyotype	FISH
45,X,t(X;14)(p22;p11).	confirmed t(X;14) - Terminal Xp, 14q deletion
45,X,t(Y;19)	Confirmed translocation
47,XY,mar	The mar is derived from chrom. 15
47, XY, +18	Trisomy 18.
46,XX, dup2q	FISH: partial trisomy 15q, del tel2q
46,XY,add(5p)	Confirmed that added material was derived from chromosome 14.
46,XY,t(4;16)	translocation 4;16 - LSI 16p13.3 : not deleted
46,XY, ? 22q	LSI 22q11.2 : deleted (DiGeorge syndrome).
46,XY, interstitial del (5q)	confirmed
47,XX, +mar	47,XX,der22 t(11;22)(q23.3;q11.2)
46,XY, del (1q43q44)	Subtel. Del of 1q
46, XY, del (7p21.1)	Confirmed
46, XY, r(11)	Ch 11p subtel. is deleted

Figures

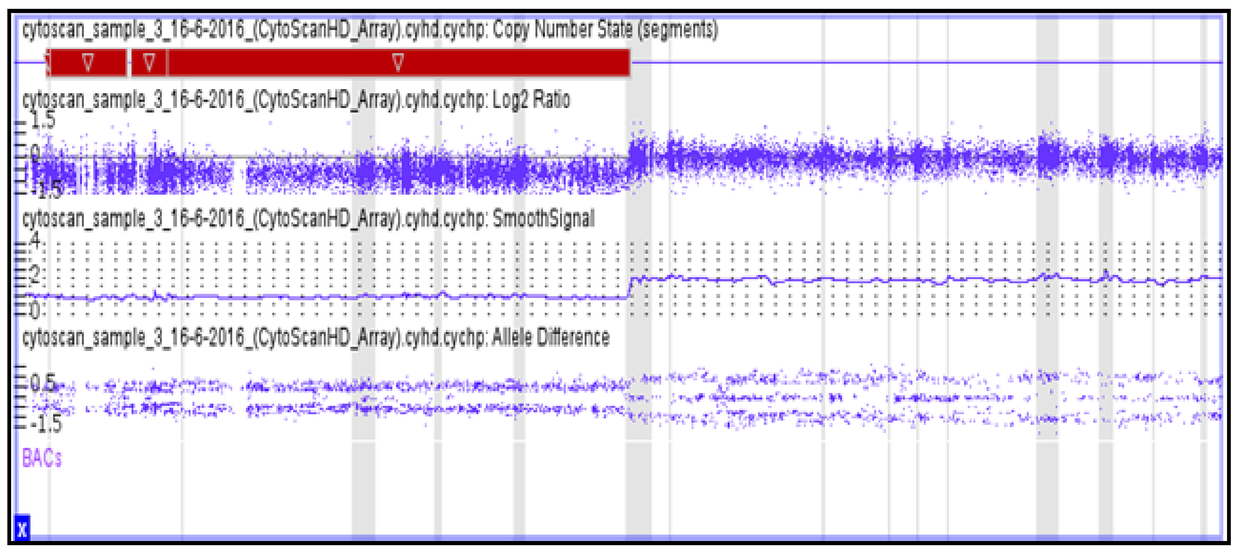




Patient with 47,XXX



ACGH of Male Patient with dysmorphic facies, undescended and retrocerebellar cyst with prominent cerebrospinal fluid spaces (CSF) spaces
Karyotype 46, XY, del (7p21.1)



Microarray for a female patient with coarse facies, microcephaly, short stature, absent labia minora and hypoplastic clitories showing **deletion in chromosome Xp (designated below the red bar) of ≈ 6 Megabase (6,228,000 bp).**

Conclusions

- The study emphasizes the **crucial need for advanced diagnostic techniques** in patients with DSD
- Improving the diagnostic strategy of such complicated disorders will be reflected on the patients and their families regarding possible **therapeutic interventions, prenatal diagnosis and genetic counseling.**

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Sex differentiation, gonads and gynaecology or sex endocrinology

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