Clinical review of 7 patients affected with 49,XXXXY syndrome

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Introduction

- 49,XXXXY polysomy, similar to Klinefleter syndrome, not the same
- Incidence ~ 1 per 85000 to 100000 male births.
- Rare condition, medical problems affecting different systems ⇒ multidisciplinary approach.

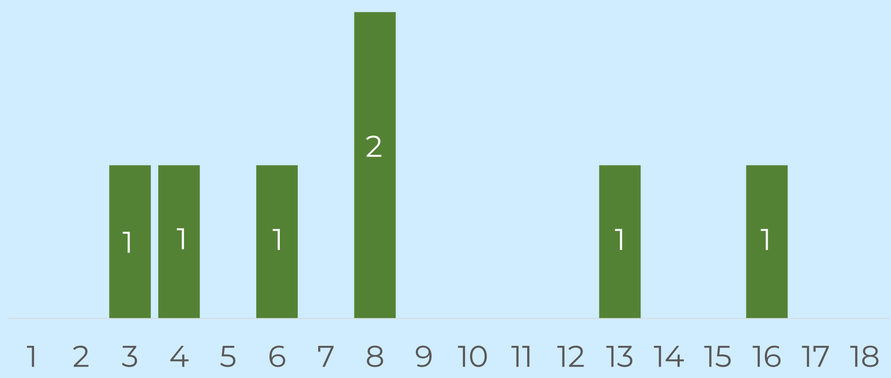
We have reviewed the clinical characteristics of patients with this anomaly from all the country who contacted the program for evaluation.

Methods

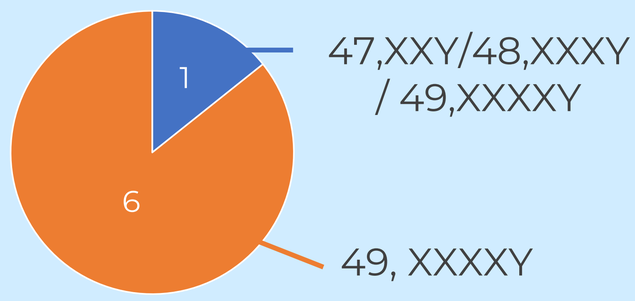
- Multidisciplinary program for the care of patients with sex chromosomal aneuploidies, started 2016
- Patients contacted trough family organizations or social networks
- Patients evaluated by an endocrinologist, psychiatrist, neuropsychologist, neurologist and clinical geneticist.

Results

Age at the evaluation



Karyotipe



Clinical features

Facial dysmorphism	
Hypertelorism	4/7
Epicanthal folds	5/7
Broad nasal bridge	5/7

Skeletal features	
Joint hyperextensibility	6/7
Congenital elbow dislocation	4/7
Clinodactyly	5/7
Pes planus	5/7
Genu valgum	4/7
Radioulnar synostosis	3/7

Cognitive development	
Attention deficit hyperactivity disorder	5/7
Anxiety behaviors	5/7
Obsessive-compulsive disorder	3/7
Impulsivity	3/7
Speech delays	7/7

Gonadal	
Low testicular size (prepubertal)	7/7
Small phallus	4/7
Cryptorchidism	4/7

All of them were shorter then their parents	
Height	-1,8 to +0,7 SD
Parents Height	-0,8 to +1,3 SD
Differential height	-2,5 to -0,6 SD

Neonatal	
Small for gestational age	4/7
Feeding difficulties	3/7
Hypotonia	7/7

Comments

- The clinical and developmental features found in these patients were similar to those previously reported, with the exception of intrauterine growth retardation.
- The diversity of clinical and developmental symptoms of this disorder make necessary a multidisciplinary approach to detect and treat early medical problems