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 through family organizations or social networks
- Patients evaluated by an endocrinologist, psychiatrist, neuropsychologist, neurologist and clinical geneticist.

Results

<table>
<thead>
<tr>
<th>Karyotype</th>
<th>47,XXY/48,XXXY</th>
<th>49,XXXXY</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at the evaluation</td>
<td>1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18</td>
<td></td>
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</tbody>
</table>

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

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

All of them were shorter than 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

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

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.