Klinefelter syndrome is the most common sex chromosome disorder (prevalence: 1/600 newborn males), characterized by at least one extra X chromosome. If the diagnosis is not made prenatally, the disorder can remain unknown until pubertal or adult age.

Kabuki syndrome is a rare, dysmorphic syndrome (1:32000 newborn) characterized by distinctive facial features, multiple anomalies and mental retardation. About 50-80% of cases are due to de novo or autosomal dominant MLL2 or KMT2B gene mutation (point mutation, deletion, duplication) located on 12q13.12; less frequently, deletions or point mutations involving the KDM6A gene, located on Xp11.3, are implicated.

Various autosomal and sex chromosome aberration have been reported in patients with Kabuki syndrome:

<table>
<thead>
<tr>
<th>Autosomal chromosome</th>
<th>Sex chromosome</th>
</tr>
</thead>
<tbody>
<tr>
<td>dup1(p13.1p22.1)</td>
<td>pericentric inversion of Y</td>
</tr>
<tr>
<td>t(3;10)(p25;p15)</td>
<td>ring Y</td>
</tr>
<tr>
<td>inv(4)(p12pter)</td>
<td>ring X</td>
</tr>
<tr>
<td>der(6)t(6;12)(q25.3;q24.31)</td>
<td>45,X</td>
</tr>
<tr>
<td>t(15;17)(15q;21q)</td>
<td>isochromosome X</td>
</tr>
<tr>
<td>dup8p22-8p23</td>
<td>pseudodicentric chromosome 13</td>
</tr>
</tbody>
</table>

**CASE REPORT**

We describe a 1-year-old boy affected by both Klinefelter and Kabuki syndromes

**Clinical history:**
He was born at 38 weeks of gestational age by cesarean section and underwent neonatal resuscitation with O₂-administration.

**At birth:**
- weight and length at the 50th percentile
- head circumference at the 35th.

Progressive postnatal weight growth retardation (associated with normal height velocity) and recurrent aspiration pneumonia needed enteral nutrition by percutaneous gastrostomy.

**Clinical features:**
- Microcephaly
- Distinctive facial appearance (big eyes with long palpebral fissures, long cilia, anteverted nostrils, extroverted lower lip with pits, cupped ears, and operated cleft palate)
- Persistence of fetal fingertip pads
- Eczema
- Congenital heart disease (atrial septal defect)
- Hypotonia
- Joint and cutaneous laxity
- Normal male genitalia
- Mild development delay

Abdominal ultrasound showed normal kidneys and liver.

**Cytogenetics:** Karyotype was 47,XXX

Sequence analysis of the Kabuki genes showed a heterozygous c.721delC de novo mutation (L241CfsX260) of the MLL2 gene.

An association between Klinefelter and Kabuki syndromes has not yet been described. These two genetic conditions casually coexist in our patient, probably due to the relatively high prevalence of the first.