Case reports

Ring chromosome 7 in a man with multiple congenital anomalies and mental retardation

Célia P Koiffermann, Aron Diamant, Deise H de Souza, Anita Wajntal

Abstract
A 39 year old male with multiple dysmorphic features was found to have an unstable ring chromosome 7. Clinical findings are presented and compared with the other five reported cases of ring chromosome 7. The main characteristics found in patients with this chromosome constitution are prenatal onset growth deficiency, bone anomalies, pigmentary or vascular skin changes, and ocular and genital anomalies.

Patients with ring chromosome 7 are rare and only five cases have been published using banding techniques.1-4 Short stature, craniofacial dysmorphism, naevus flammeus, and café au lait spots are the main characteristics observed. Two patients had mental retardation and three had intelligence within normal limits. Here we report the sixth case of ring chromosome 7 in a 39 year old man with mental retardation and multiple malformation syndrome (MCA/MR).

Case report
The proband was the product of a term pregnancy born to normal, non-consanguineous parents; the mother was 30 years old and the father 32. Birth weight was 2000 g and length was reported to be below normal. He was noted to have microcephaly, a short left palpebral fissure, a left inguinal hernia, and hypospadias. Growth and development were delayed; he walked at 24 months and his speech has always been incomprehensible. His seven sibs are normal. On physical examination when he was 39 years old (fig 1) there was a gerodermic facies, short stature (height 130 cm, less than the 3rd centile), a small, brachycephalic head (head circumference 46 cm, less than the 2nd centile), haemangiomas on the forehead and neck, ptosis of the right eyelid, apparent hypertelorism (inner canthal distance 2·5 cm, outer canthal distance 8·5 cm), upward slanting eyebrows, a beaked nose with hypoplastic nares, high set ears, thin lips, long philtrum, malar hypoplasia, kyphoscoliosis, low set and widely spaced nipples, and pigmented naevi, mainly on the back and abdomen. There was a repaired left inguinal hernia, small penis with hypospadias, and enlarged scrotum. The hands were small and broad with bilateral shortening of the second phalanges of the fifth fingers, more evident on the left. The feet were small with bilateral partial syndactyly of the phalanges of the third and fourth toes. He had severe mental retardation, dysarthric speech, and urinary incontinence at night.

Dermatoglyphics showed the following features. Left hand: L° (17), L® (15), L° (15), W° (10), c® (10). Mainline formula: 7·5°·5°·4·13-t-L®·O.O.O.L. Right hand: L° (19), L° (15), L° (15), W° (12), W° (14). Mainline formula: 9·X°·5°·4·13-t-L®·O.O.O.L.

Further tests on the proband were refused.

Genetic Counselling Unit, Department of Biology, Institute of Biosciences, University of São Paulo, SP, Brazil.
C P Koiffermann, D H de Souza, A Wajntal

Service of Child Neurology, Department of Neurology, Faculty of Medicine, University of São Paulo, SP, Brazil.
A Diamant

Correspondence to Dr Koiffermann, Unidade de Aconselhamento Genético, Departamento de Biologia, Instituto de Biociências, Universidade de São Paulo, Caixa Postal 11.461, CEP 05499, São Paulo, Brazil.

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CYTOGENETIC FINDINGS
Chromosome analysis was performed on peripheral blood lymphocytes using GTG and CBG banding. Fifty cells were analysed, all with at least one ring chromosome 7 (fig 2). The length of this ring chromosome in the majority of cells was equivalent to the length of a chromosome 7 with breakpoints in the terminal regions of both arms. In some cells the ring was of reduced size or double sized with two centromeric regions. Cells with two single or double rings were also found, but there were no cells without
the ring chromosome. The mother and one brother have normal karyotypes. The father refused cytogenetic investigation.

Discussion
To our knowledge there are only five previously reported patients with ring chromosome 7. Prenatal onset growth deficiency, craniofacial dysmorphism, and skin lesions have been described as the main characteristics in patients with this chromosome constitution (table). Bone anomalies were also present in all patients described. Ocular anomalies in three out of four patients and genital anomalies in two out of five boys were also observed and therefore must be included as phenotypic characteristics present in patients with ring chromosome 7.

The bone anomalies present in these patients are...
Main features observed in patients with ring chromosome 7 (six cases).

<table>
<thead>
<tr>
<th></th>
<th>Zackai and Breg</th>
<th>Nakano and Miyamoto</th>
<th>DeLozier et al</th>
<th>Barros et al</th>
<th>Present case</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age first examined</strong></td>
<td>23 mth</td>
<td>16 mth</td>
<td>4 y</td>
<td>8 ± 12 y</td>
<td>14 mth</td>
</tr>
<tr>
<td><strong>Sex</strong></td>
<td>M</td>
<td>M</td>
<td>M</td>
<td>F</td>
<td>M</td>
</tr>
<tr>
<td><strong>Birth weight (g)</strong></td>
<td>2800</td>
<td>2200</td>
<td>1160</td>
<td>2650</td>
<td>2350</td>
</tr>
<tr>
<td><strong>Birth length (cm)</strong></td>
<td>0</td>
<td>0</td>
<td>30.2</td>
<td>46</td>
<td>45</td>
</tr>
<tr>
<td><strong>Maternal age at birth of proband</strong></td>
<td>35</td>
<td>32</td>
<td>0</td>
<td>28</td>
<td>29</td>
</tr>
<tr>
<td><strong>Paternal age at birth of proband</strong></td>
<td>34</td>
<td>34</td>
<td>0</td>
<td>34</td>
<td>38</td>
</tr>
<tr>
<td><strong>Growth pattern</strong></td>
<td>&lt;3rd centile</td>
<td>&lt;3rd centile</td>
<td>&lt;3rd centile</td>
<td>10th centile</td>
<td>&lt;3rd centile</td>
</tr>
<tr>
<td><strong>Mental retardation</strong></td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td><strong>Dysarthric speech</strong></td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td><strong>Microcephaly</strong></td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td><strong>Ocular anomalies</strong></td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td><strong>Skin lesions</strong></td>
<td>+</td>
<td>+</td>
<td>+</td>
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<td>+</td>
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<tr>
<td><strong>Bone anomalies</strong></td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td><strong>Genital anomalies</strong></td>
<td>+</td>
<td>-</td>
<td>0</td>
<td>-</td>
<td>0</td>
</tr>
</tbody>
</table>

*Data from DeLozier et al* and Schinzel. 1
0 = not reported by the authors.

Some is present in the original ring. This characteristic is also present in most of the chromosome instability syndromes, such as Bloom’s syndrome, and may be secondary to ring chromosome instability. Major abnormalities are probably secondary to deletions or duplications of specific genetic material affecting fetal development.

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