LETTERS TO THE EDITOR

A new form of infantile spinal muscular atrophy

We read with great interest the article by Borochowitz et al.2 describing a possible new form of infantile spinal muscular atrophy (SMA) characterised by congenital contractions, multiple bone fractures, and early death.

A few years ago we performed a detailed study of the skeletal changes in the long bones in 11 infants with neuromuscular diseases.1 SMA was diagnosed at necropsy in six of these infants and these infants were clinically similar to the two patients reported by Borochowitz et al.1 The infants with SMA in our study were from four families without parental consanguinity. The first three cases were consecutive offspring (two females and one male) of healthy parents, which provides additional support for the suggestion of Borochowitz et al.2 that this is an autosomal recessive disorder. The consistent radiographic findings of each infant with neuromuscular disease in our study were thin, hypomineralised, and elongated long bones. In addition, all infants with SMA showed multiple diaphyseal or metaphyseal fractures or both, primarily involving the long bones of the upper extremities. A morphometric evaluation of the long bones showed a more pronounced decrease in periosteal diameter, cortical thickness, and cortical area in the patients with SMA as compared to infants with other types of neuromuscular disease, for example, congenital myotonic dystrophy.3 Since the shape and growth of bones are determined by an interactive process between the intrinsic properties of bone and biomechanical forces, severe fetal immobility can lead to hypoplastic bones and fragility. Moreover, stiff joints may play a role in the genesis of the fractures, owing to diminished absorption of the mechanical forces applied to the fragile bones during delivery and postnatal handling of the newborn.4 Although in our experience joint contractures and bone fractures are non-specific findings, since they are related to the duration and severity of the intrauterine akenisia, their presence in infants with SMA probably implies a severe disorder of intrauterine onset. This assumption is further supported by the fact that many of the infants studied also had other abnormalities associated with fetal akenisia, such as polyhydramnios, intrauterine growth retardation, microgastria, and pulmonic hypoplasia.5 In conclusion, our previous studies support the suggestion of Borochowitz et al.2 of a new form of infantile SMA distinct from the common classical form of SMA I.

A GARCIA-ALIx
J R RODRIGUEZ
J QUREKO
Division of Néumatology,
Hospital Infantil La Paz,
Paseo de la Castellana 261,
Madrid 28046,
Spain.


MASA syndrome (a form of complicated spastic paraplegia) and X linked hydrocephalus: variable expression of the same mutation at Xq28? Call for families

Spastic paraplegia is a common autosomal dominant disorder. In the complicated form, it may be present in combination with mental retardation or ophthalmologic anomalies or both.1 In the X linked recessive 'MASA' syndrome, the Spastic paraplegia is combined with Mental retardation, Aphasia (late speech development), and Adducted thumbs.2 We had the opportunity to examine two families where several males had either MASA syndrome or congenital hydrocephalus. Extensive data on both families have been studied also had other abnormalities associated with fetal akenisia, such as polyhydramnios, intrauterine growth retardation, microgastria, and pulmonic hypoplasia. In conclusion, our previous studies support the suggestion of Borochowitz et al. of a new form of infantile SMA distinct from the common classical form of SMA I.

S M. Rodriguez, J I Garcia-Alix, A Palacios, J Paniagua
Departamento de Neumología, Hospital Infantil La Paz, Madrid, Spain.


Clinical comparison between male patients with MASA syndrome and mentally retarded males from families with X linked hydrocephalus.

<table>
<thead>
<tr>
<th>Clinical features</th>
<th>MASA males&lt;sup&gt;2,4&lt;/sup&gt; (n = 22)</th>
<th>XLM males&lt;sup&gt;1,3&lt;/sup&gt; (n = 20)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Head circumference (cm)</td>
<td>100-98</td>
<td>100-98</td>
</tr>
<tr>
<td>Mental development (IQ)</td>
<td>40-75</td>
<td>20-50</td>
</tr>
<tr>
<td>Adducted thumbs</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>Spastic paraplegia (hyperreflexia)</td>
<td>17/17</td>
<td>19/20</td>
</tr>
<tr>
<td>Abnormal CT scan of the brain (enlargement of the ventricles)</td>
<td>2/3</td>
<td>5/7</td>
</tr>
</tbody>
</table>

Clinical features of MASA males<sup>2,4</sup> and XLM males<sup>1,3</sup> indicate a significant difference in head circumference, mental development, and adducted thumbs. Spastic paraplegia and abnormal CT scan of the brain are more common in MASA males. However, the number of cases is small, and further studies are needed to confirm these findings.
A new form of infantile spinal muscular atrophy.

A García-Alix, J I Rodriguez and J Quero

doi: 10.1136/jmg.29.3.215

Updated information and services can be found at:
http://jmg.bmj.com/content/29/3/215.1.citation

*Email alerting service*

*These include:*

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

*Notes*

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/