patients. Skin biopsies were performed in 8.2% of cases; this was lower than the rate reported by Davila¹ (20%) but higher than the rate described by Adısen (4.4%).²

In our experience, common dermatologic diseases are often not correctly diagnosed by physicians from other specialties. In addition, there is room for improvement in the formal description and in the differential diagnosis of skin diseases. Expert dermatologic assessment usually facilitates inpatient diagnosis and management. Better training should be considered for medical students and residents and possibly even for medical staff in other specialties.

Bibliografía


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Frontal congenital lipoma and lipoma of the corpus callosum in an infant: A case report

Lipoma frontal congénito y lipoma del cuerpo caloso en un lactante: Informe de un caso

To the Editor,

An otherwise healthy 4-month-old girl who had been born full-term without birth trauma or prenatal or neonatal complications was brought to our practice because of a frontal tumor that had been present since birth. Physical examination revealed a deep frontal tumor of medium consistency that was mobile, unattached to the deeper layers, and without epidermal changes (Fig. 1). The rest of the examination was normal. No hypertelorism, nasal alterations, or dysmorphic facial features were observed.

A soft-tissue cranial ultrasound performed when the infant was 2 days old showed slight thickening of the subcutaneous tissue; this was also visible in a second ultrasound performed 2 months later. The diagnosis was congenital frontal lipoma.

The patient was lost to follow-up but returned when she was 8 months old. A brain magnetic resonance imaging (MRI) study showed an interhemispheric hyperintense mass on both T1- and T2-weighted sequences and a hypointense mass

Table 3 Dermatologist’s diagnosis.

<table>
<thead>
<tr>
<th>Skin disease</th>
<th>n (%)</th>
<th>Skin disease</th>
<th>n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Contact dermatitis</td>
<td>39 (9.4)</td>
<td>Psoriasis</td>
<td>9 (2.2)</td>
</tr>
<tr>
<td>Fungal infections</td>
<td>35 (8.4)</td>
<td>Recurrent aphthous stomatitis</td>
<td>8 (1.9)</td>
</tr>
<tr>
<td>Drug reactions</td>
<td>28 (6.7)</td>
<td>No dermatologic disease</td>
<td>8 (1.9)</td>
</tr>
<tr>
<td>Cellulitis</td>
<td>27 (6.4)</td>
<td>Seborrheic dermatitis</td>
<td>7 (1.7)</td>
</tr>
<tr>
<td>Xerosis cutis</td>
<td>15 (3.6)</td>
<td>Behçet disease</td>
<td>7 (1.7)</td>
</tr>
<tr>
<td>Pruritus</td>
<td>15 (3.6)</td>
<td>Malignancy</td>
<td>7 (1.7)</td>
</tr>
<tr>
<td>Vasculitis</td>
<td>14 (3.4)</td>
<td>Oral candidiasis</td>
<td>7 (1.7)</td>
</tr>
<tr>
<td>Stasis dermatitis</td>
<td>14 (3.4)</td>
<td>Acneiform disease</td>
<td>6 (1.4)</td>
</tr>
<tr>
<td>Herpes labialis</td>
<td>13 (3.1)</td>
<td>Rosacea</td>
<td>5 (1.2)</td>
</tr>
<tr>
<td>Infections (folliculitis,</td>
<td>13 (3.1)</td>
<td>Traumatic ulcer</td>
<td>5 (1.2)</td>
</tr>
<tr>
<td>furunculosis, paronychia)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Neurodermatitis</td>
<td>12 (2.9)</td>
<td>Spontaneous and traumatic ecchymosis</td>
<td>5 (1.2)</td>
</tr>
<tr>
<td>Urticaria</td>
<td>11 (2.6)</td>
<td>Vascular disease (thrombophlebitis, ischemia)</td>
<td>5 (1.2)</td>
</tr>
<tr>
<td>Pressure sore</td>
<td>11 (2.6)</td>
<td>Autoimmune bullous disease</td>
<td>4 (1.0)</td>
</tr>
<tr>
<td>Intertrigo</td>
<td>11 (2.6)</td>
<td>Seborrheic keratosis</td>
<td>4 (1.0)</td>
</tr>
<tr>
<td>Herpes zoster</td>
<td>10 (2.4)</td>
<td>Radiodermatitis</td>
<td>3 (0.7)</td>
</tr>
<tr>
<td>Miscellaneous</td>
<td>59 (14.1)</td>
<td>Total</td>
<td>417</td>
</tr>
</tbody>
</table>
Furthermore, intracranial lipomas are also rare, accounting for just 0.06–0.46% of intracranial lesions. Most are located in the midline/interhemispheric region, most often in the corpus callosum. In about 50% of cases other disturbances, frequently associated with varying degrees of hypoplasia or agenesis of the corpus callosum, are identified in the surrounding nervous structures.

Subcutaneous lipomas in association with intracranial lipomas are even rarer. The association could be related to the abnormal migration and proliferation of neural crest cells. Abnormal neural crest development results in many craniofacial malformations, known as neurocristopathies, including facial midline clefts. Intracranial and extracranial lipomas may be independent entities or connected through a frontal bone defect on the skull.

Frontonasal dysplasia (FND) is a developmental alteration of the craniofacial region that comprises a spectrum of anomalies of the frontonasal area, including hypertelorism, nasal anomalies, and/or lip-palate cleft. The exact origin of FND is unknown and most cases are sporadic, although a mutation in the TGF2 gene has been observed in familial cases of FND, which are very rare.

Patients with FND may present with hypoplasia or agenesis of the corpus callosum and/or a corpus callosum lipoma. In a case series of patients with FND, all 8 patients had lipoma of the corpus callosum. Markers strongly associated with FND are falx cerebri calcifications and extracranial lipomas.

Midline lipomas of the face and other craniofacial anomalies may be associated with intracranial malformations, including intracranial lipomas.

Brain MRI for the study of intracranial structures combined with clinical follow-up to monitor neurological changes seems to be the gold standard.

Pai syndrome should be included in the differential diagnosis of FND spectrum anomalies. This syndrome consists of pericallosal lipomas associated with facial abnormalities such as cutaneous polyps of the face and nasal mucosa.

Figure 1  A soft, mobile, asymptomatic nodule in the frontal midline area of a 4-month-old girl.

Figure 2  Interhemispheric hyperintense structure on T1-weighted image consistent with a lipoma associated with hypoplasia of the splenium of the corpus callosum (bright fat tissue on T1 sequence). (A) Sagittal plane. (B) Transverse/axial plane.
midline cleft, and midline pericallosal lipoma.\textsuperscript{10} As with our patient, a nasal fibroscopy should be performed to rule out this syndrome.

Although the majority of patients with intracranial lipomas are asymptomatic,\textsuperscript{10,11} a small number of patients may present neurological symptoms such as seizures, headache, and/or behavioral or psychosocial disorders.\textsuperscript{8} Routine neurosurgical treatment is not recommended because the surgical risk usually outweighs the benefits of the intervention.\textsuperscript{4} Surgical resolution of extracranial lipoma may provide cosmetic improvement and better quality of life.

The prognosis and psychomotor development of patients with intracranial lipomas is not clear, but based on data from patients with FND and Pai syndrome, their prognosis would appear to be favorable, with normal psychomotor development and no neurological impairment.\textsuperscript{8,10} Some patients with FND may have psychological alterations such as misanthropy and shyness.\textsuperscript{8,9}

Lipomas are rare in children and are even rarer at birth. Facial midline lipomas should be assessed by a multidisciplinary team consisting of a dermatologist, neurosurgeons, an otorlaryngologist, and radiologists. Neurologic images should be taken and in cases associated with corpus callosum or pericallosal lipoma, FND and Pai syndrome must be ruled out. Whether our patient represents an isolated case of frontal congenital lipoma with associated cerebral lipoma or an incomplete case within the spectrum of FND is currently unknown.

Bibliografía


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Rapamicina tópica al 0,2% para el tratamiento de angiofibromas faciales y máculas hipomelanóticas en la esclerosis tuberosa

Topical 0.2% Rapamycin to Treat Facial Angiofibromas and Hypomelanotic Macules in Tuberous Sclerosis

La esclerosis tuberosa (ET) es un trastorno neurocutáneo de herencia autosómica dominante causado por mutaciones en el gen supresor tumoral, TSC1 (cromosoma 9q34) o TSC2 (cromosoma 16p13.3), que codifican para las proteínas hamartina y tuberina, respectivamente. Dichas proteínas son responsables de formar un complejo que inhibe la molécula diana de la rapamicina en mamíferos (mTOR), fundamental en la regulación del ciclo celular. El resultado es una proliferación celular descontrolada, caracterizándose por la aparición de hamartomas en múltiples órganos, incluyendo el piel, el riñón y el sistema nervioso central.\textsuperscript{1,2} Rapamicina (sirolimus) es un fármaco inmunosupresor que actúa mediante la inhibición de mTOR. Se ha utilizado clásicamente por vía oral para evitar el rechazo en el trasplante renal, dadas sus propiedades antineoplásicas al inhibir la neoangiogénesis y la proliferación de células tumorales, y se ha comprobado su eficacia en la disminución del número y tamaño de los tumores en los pacientes diagnosticados de ET. Recientemente se ha publicado la utilidad de rapamicina tópica para el tratamiento de angiofibromas faciales\textsuperscript{3-7} y en la reducción de máculas hipomelanóticas\textsuperscript{8} en pacientes con ET.

Presentamos el caso de un paciente, varón de 13 años de edad, diagnosticado clínicamente de ET a los 4 meses por presentar un cuadro con varias manifestaciones características de la enfermedad: epilipsia, múltiples máculas hipomelanóticas y angiofibromas faciales. El estudio genético confirmó la presencia de ET esporádica, debido a una mutación c5043C>G en el exón 38 del gen TSC2, que modifica la secuencia de la proteína p.N1681K. La resonancia magnética nuclear cerebral demostró la presencia de múltiples...