Case Report

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Basal Cell Nevus Syndrome and Congenital Hydrocephaly

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Key Words

Basal cell nevus syndrome
Congenital hydrocephaly

Abstract

A case of basal cell nevus syndrome or Gorlin's syndrome is reported in a new-born. The skin condition is associated with congenital hydrocephaly and skeletal malformations. To our knowledge, this is the first case of basal cell nevus syndrome with skin tumors present at birth and localized on the fingers

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Basal cell nevus syndrome is an autosomal dominant inherited condition although some sporadic cases have been reported [1].

It was first described by Gorlin and Goltz in 1960 [2] and characterized by five major components: multiple basal cell carcinomas appearing at an early age; jaw cysts; skeletal anomalies, especially of the ribs, spine, skull and metacar-pals; soft tissue calcification, and palmar and plantar pits. A variety of other, less common defects have been described [3], among which are neurologic abnormalities.

We saw a 5-month-old boy with basal cell nevus syndrome, associated with congenital hydrocephaly. Skin lesions were present since birth and localized on the fingers, whereas they usually appear at puberty [4] and arise on the face and trunk.

smooth-surfaced rounded elevated papules, flesh-colored or pig-mented and varying in size from 1 to 6 mm in diameter, localized on the lateral sides of the fingers of both hands (fig. 1). Pits are absent. Further clinical examination reveals a particular facies with ocular hypertelorism, frontal bossing (fig. 2) and macrocephaly.

Routine laboratory investigations were within normal limits. The karyotype is normal. A skin biopsy is performed, and histological as well as ultrastructural examination confirmed the presence of a superficial and middermal nodule, which consists of basophilic cells which are arranged in characteristic palisades (fig. 3). This confirms the diagnosis of basal cell carcinoma. Ophthalmological examination cannot reveal any anomaly.

A radiological checkup shows a fourth bifid rib on the right side and a spina bifida occulta (D2-D6). There are no jaw cysts, no calcification of the falx cerebri nor brachymetacarpalism.

Discussion

Case Report

This male infant is the first born from nonconsanguineous parents.

The family history reveals the death of an uncle on the father's side at the age of 5 years from an intracranial sarcoma.

At the age of 5 months, he is admitted to our hospital because of an evolutive macrocephaly.

Neuroimaging studies show a triventricular hydrocephaly due to aqueduct stenosis and a diffusely hypertrophic cerebellum. A ventri-culoperitoneal drain is placed.

He is presented at our consultation for skin lesions present since birth and slowly increasing in number. The individual lesions are

The diagnosis of Gorlin's syndrome or basal cell nevus syndrome is retained because of the presence of congenital hydrocephaly, basocellular nevi on the hands and costover-tebral malformations.

This genodermatosis, which progresses during a whole lifetime, is inherited in an autosomal dominant fashion, but sporadic cases have been reported.

This case is particularly interesting because of the early skin manifestations and their unusual localization. In Gorlin's syndrome, basocellular nevi indeed usually appear during puberty. They occur most frequently on the face,

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Fig. 1. Basal cell nevus syndrome. Congenital papulonodular lesions on fingers.

Fig. 2. Basal cell nevus syndrome. Typical facies with hypertelorism and frontal bossing.

neck, trunk and armpit. Nevertheless, congenital lesions and lesions localized on limbs have been described [4].

In infancy the prognosis of basal cell nevus syndrome is essentially determined by the associated neurologic disorders, which necessitate a complete investigation.

Congenital hydrocephaly is not frequent; it has been described by Scully and Gorlin in 1965 and confirmed by Lycka and Chichak in 1985.

Other abnormalities can also be present including oligophrenia, schizophrenia, agenesis of the corpus callosum, degenerative pyramidal syndrome and especially intracranial tumors, more particularly medulloblastomas, responsible for the mortality risk during infancy.

Cutaneous and neurologic follow-up examinations are warranted in order to detect possible degenerative changes in skin tumors or neurologic dysfunction.

Neuroimaging studies may predict intracranial involvement.

Fig. 3. Biopsy of skin tumor. Basal cell carcinoma.

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