

OCULAR MANIFESTATIONS IN PROTEUS SYNDROME

MANIFESTACIONES OCULARES EN EL SÍNDROME DE PROTEUS

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ABSTRACT

Case report: Congenital disfiguring malformations are rare and usually have a multifactorial aetiology. Here we report on the ocular manifestations seen in a patient with Proteus syndrome. The retina showed retinal dysgenesis, retinal pigmentary abnormalities and optic nerve hypoplasia. Other abnormalities included strabismus and high myopia.

Discussion: Proteus syndrome is a complex hamartomatous disorder defined by local overgrowth, subcutaneous tumours and various bone, cutaneous and/or vascular anomalies. The incidence of ocular malformations in Proteus syndrome is unknown, however a meticulous cranio-facial examination and a systematic study of the eye is required to improve the medical care of these patients (*Arch Soc Esp Oftalmol* 2007; 82: 175-178).

Key words: Proteus syndrome, retinal dysgenesis, myopia, retinal pigmentary abnormalities, optic nerve hypoplasia.

RESUMEN

Caso clínico: Las malformaciones congénitas deformantes son raras y tienen una etiología multifactorial. Presentamos las manifestaciones oculares de un caso clínico de Síndrome de Proteus. La retina mostraba una desorganización difusa, alteraciones pigmentarias e hipoplasia de nervio óptico. Otras alteraciones eran estrabismo y alta miopía.

Discusión: El Síndrome de Proteus es un complejo trastorno hamartomatoso caracterizado por un crecimiento local exagerado, tumores subcutáneos y diversas malformaciones óseas, cutáneas y/o vasculares. La incidencia de las malformaciones oculares en el Síndrome de Proteus es desconocida, precisando un examen craneofacial minucioso y un estudio sistemático ocular en estrecha relación multidisciplinaria para mejorar la asistencia de estos pacientes.

Palabras clave: Síndrome de Proteus, disgenesia retiniana, miopía, anomalías pigmentarias retinianas, hipoplasia de nervio óptico.

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CASE REPORT

A female born within term via vaginal labor, the first child of parents without consanguinity. In week 21 of gestation, a control echography revealed a slight bilateral ventriculomegalia which persisted in successive controls. No other events were observed during the pregnancy. The Apgar test score was 3/10, 4/10 and 8/10 at 1, 10 and 20 minutes respectively, requiring endotracheal intubation during labor. The weight at birth was 3,480 grammes, height 55 cm and cranial perimeter 39.5 cm. At birth, the baby exhibited dysmorphic features: rough fascia, front-facing narines, hypertelorism, microretrognathia and frontal asymmetry with left prominence. General hypotony progressing towards general hypertony with sharp reflexes, clonus and neonatal period. An EEG was performed, which showed a discrete diffuse lentification. The brain echography revealed asymmetric ventriculomegalia with left side predominance and hemispheric asymmetry towards the left side. The cranial CAT scan showed a prominence of the ventricular system. The NMR revealed cortical dysplasia with nodular heterotopy areas (figs. 1 and 2).

The metabolic and infection screening produced normal results. Chariotype 46XX.

At 7 months the baby was referred to the ophthalmology service due to 15° esotropy in the right eye. She exhibited left eye dominance, nistagmus in abduction in both eyes. The retina appeared dystrophic with diffuse disorganization and retinal pigmentary abnormalities. Optic nerve hypoplasia (fig. 3).

The patient exhibited a myopia of 13.00 dioptres (2 to 90°) in right eye and 14.00 dioptres (2 to 180°) in left eye. Optical correction was prescribed. The visual evoked potentials were pathological with increased latency period. An electroretinogram could not be carried out due to lack of cooperation. At 14 months, the left hemicranial hypertrophy prevented the use of eyeglasses. Contact lenses were prescribed, which were well tolerated (fig. 4).

At 10 months, the patient exhibited an important psychological and motor retard, hypertonia of limbs and cervicoaxial hypotony. There was no cephalic control or sedestation. Visual contact improved with greater interaction with the environment and social smile. Electroencephalographic controls showed interhemispheric asymmetry, without irritating paroxysm. The patient exhibited a progressive increase of the left hemibody, plagiocephalia and lowering of the ipsilateral orbit (fig. 5).



Fig. 1: Asymmetric ventriculomegalia with left side predominance.

Progressive appearance of deformities: tibia vara, coxa valga and bilateral femoral thinning.

At 15 months, the patient exhibited macrocephalia with involvement of the craneofacial area. Epidermic nevus and dispersed hemangiomas with left side predominance. Spasticity as response to stimuli. At 16 months, a tracheostomy due to laringo tracheomalacia was performed. The patient died six days postop due to pneumonia.

DISCUSSION

The above described case is an early and severe Proteus syndrome with a negative overall evolution due to convulsions from the neonatal period, laringotracheomalacia, and a final decease due to pneumonia, a cause of death described by Cohen Jr (1). The syndrome is hereditary but the diagnosis is usually late and suspected due to exaggerated and progressive hypergrowth of any bodily region, which usually stops after puberty. Over 200 cases of



Fig. 2: Prominence of the ventricular system towards the left side.

this syndrome have been published in the literature, but Spanish literature has not given a great deal of attention to it and at least three cases have been published.

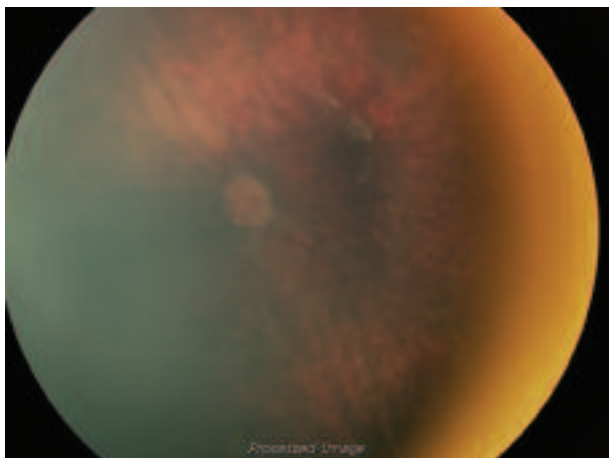


Fig. 3: Diffuse disorganization and retinal pigmentary abnormalities. Optic nerve hypoplasia.

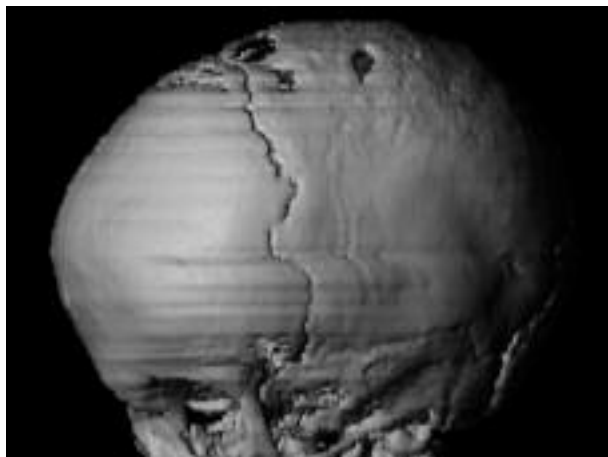


Fig. 4: Large left hemicranial hypertrophy.

The rate of faulty diagnosis is high because, although the diagnostic criteria have been published (2), their application has not been consistent. In addition, their publication came only after many cases were reported.

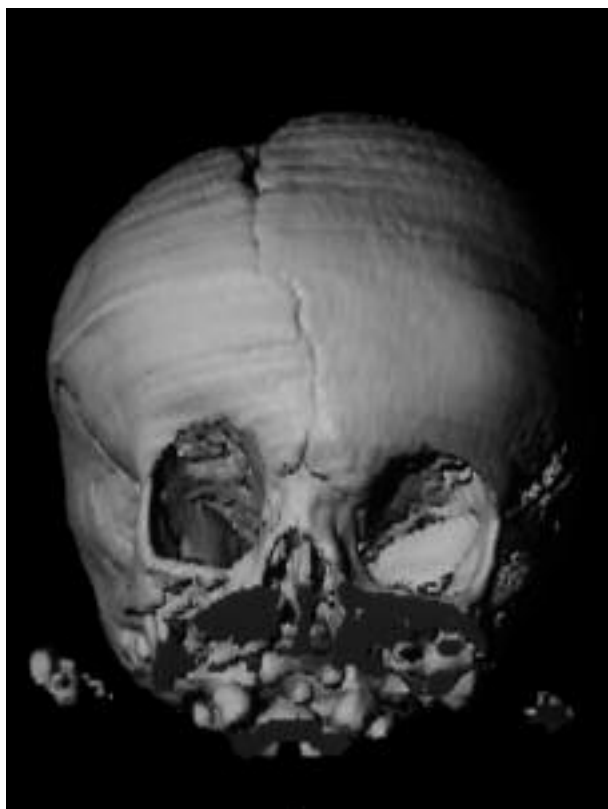


Fig. 5: Age ten months. Plagiocephalia. Descent of the ipsilateral orbit.

A differential diagnostic must be made vis-à-vis other disorders (3), with neuroectodermic mosaic phenotypes such as the epidermic nevus syndrome, cranial cutaneous lipomatosis, Delleman syndrome, Schimmelpenning syndrome, Goltz syndrome and Goldenhar syndrome, among others.

The prognosis is usually correlated to the severity of neurological involvement, skeletal deformities, internal lipomatosis and neoplastic appearances. De Becker et al (4) described in 2000 the ocular expressions of the syndrome, with the most frequent being strabismus and epibulbar tumors. Similarly, Sheard et al (5) described a new ophthalmic expression which included, in addition to the above described cataract and myopia, other abnormalities such as keratopathy, vitreous structure anomalies, chorioretinal hamartoma associated to retinal serous detachment and vitreous hemorrhage. On many occasions, a systematic ophthalmological study is lacking. Likewise, many publications do

not make reference to ocular expressions. All this combined leads to a faulty assessment of the prevalence of ophthalmological expressions.

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