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SHORT COMMUNICATION

Case report

Retinal detachment in focal dermal hypoplasia

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BACKGROUND. *Focal dermal hypoplasia is a systemic disease that includes well recognized ocular abnormalities. Retinal detachment has not previously been reported as a part of this syndrome.*
PURPOSE. *To report a case of focal dermal hypoplasia with an associated retinal detachment.*
METHODS. *Single case report of a child with a colobomatous retinal detachment and the focal dermal hypoplasia syndrome.*
CONCLUSIONS. *Retinal detachments may occur as part of the focal dermal hypoplasia syndrome. Expanding knowledge of this syndrome may help ophthalmologists diagnose this rare condition. (Eur J Ophthalmol 2004; 14: 166-8)*

KEY WORDS. *Focal dermal hypoplasia, Goltz syndrome, Goltz-Gorlin syndrome*

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Focal dermal hypoplasia is an X-linked dominant disorder that presents only in females, as males die in utero. The disease, known also as either Goltz or Goltz-Gorlin syndrome, manifests with typical features of the skin, hands and feet, mouth and eyes (1, 2). The most typical features of the disease involve the skin and include dermal atrophy, linear pigmentation of the skin, herniation of fat through the dermal defects, and mucous membrane papillomas. Anomalies of the hands and feet are common and include syndactly, polydactly and camptodactly. Anomalies of the mouth include hypoplasia of the teeth. Ocular abnormalities are a consistent finding in this syndrome and include strabismus, microphthalmia, ectopia lentis, and colobomas of the iris and choroid (3). We present a patient with a colobomatous retinal detachment, a finding not previously associated with this condition.

Case report

A fourteen month old female infant who was diagnosed at birth with Goltz syndrome was referred to our pediatric retina service for evaluation of a possible retinal detachment. The child was born at 36 weeks of age and weighed 2300 grams. The child demonstrated several characteristic features of Goltz syndrome including the typical skin changes (Fig. 1), mal-

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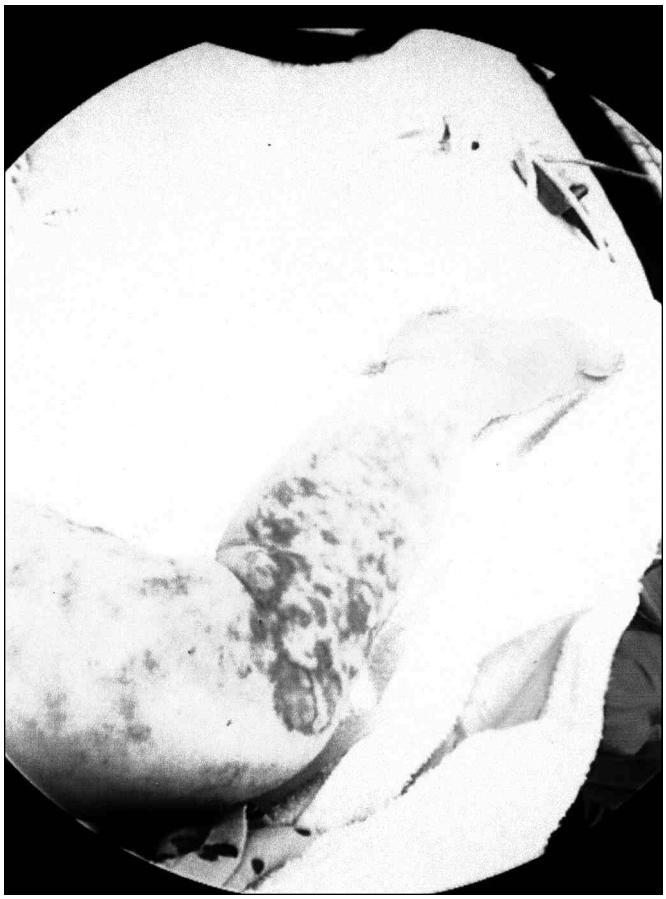

Fig. 1 - *Skin changes showing dermal atrophy.*

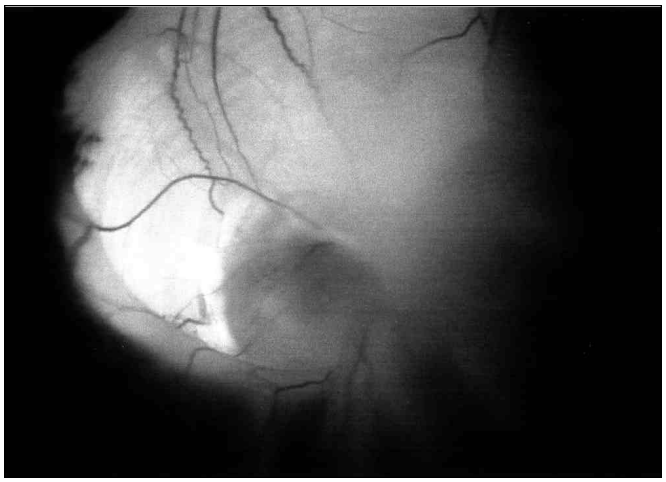

Fig. 2 - *Choroidal coloboma with retinal detachment in open funnel configuration of right eye.*

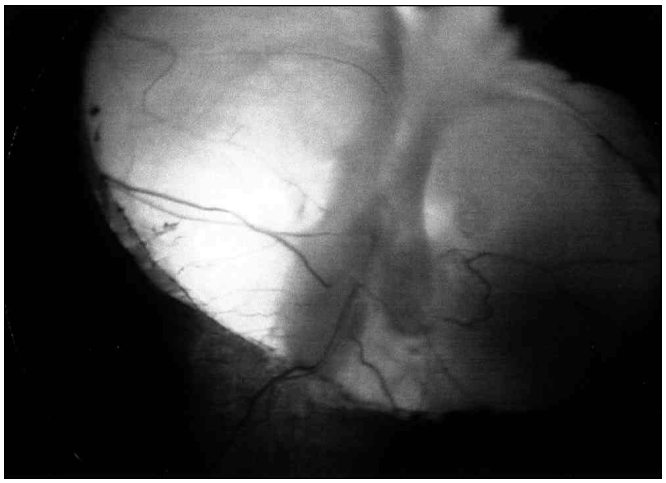

Fig. 3 - *Choroidal coloboma without retinal detachment left eye.*

formations of the hands and feet, dental hypoplasia, and developmental delay. The patient illustrated a blink response to light in both eyes, and was evaluated further during an exam under anesthesia.

The patient was mildly microphthalmic in both eyes with corneal diameters of 8.5 mm. The eyelids and lashes had a normal appearance. Both eyes had inferior iris colobomas. The right eye was noted to have a large choroidal coloboma centered on the optic nerve and involving the posterior pole. The retina was entirely detached in an open funnel configuration (Fig. 2). The left eye demonstrated a choroidal coloboma without retinal detachment (Fig. 3).

The retinal detachment was repaired utilizing intravitreal autologous plasmin enzyme (APE). A pars plana approach was performed to remove the central vitreous. Bimanual membrane peeling allowed for a complete separation of the posterior hyaloid. The freely mobile retina was then settled using perfluorocarbon, and examined extensively for the presence of a peripheral retinal break.

The perfluorocarbon was then removed and the crater-like surface of the coloboma was extensively searched. No break was identified; therefore a posterior retinotomy was created to allow for the subsequent internal drainage of the detachment under air. When the retina was completely flat, laser retinopexy was applied to the retinotomy site, and silicone oil was used to fill the eye.

DISCUSSION

Focal dermal hypoplasia is a rare X-linked dominant syndrome. Patients afflicted with this disease manifest typical ophthalmic features including colobomas of the iris and choroids. Our patient presented with a complete retinal detachment in the right eye. Retinal detachments have not previously been associated with this disorder, and a direct causal connection of the retinal detachment and this rare disease may not exist. Knowledge of this syndrome is germane for ophthalmologists who may be consulted to help confirm a diagnosis of focal dermal hypoplasia.

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