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Sir,

Poland anomaly associated with ipsilateral combined hamartoma of retina and retinal pigment epithelium

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Poland anomaly appears with an incidence of about 1 in 30 000 live births and is three times more common in male than in female subjects. The syndrome was first described in 1841 by Alfred Poland, a British ophthalmologist, and typically comprises the combination of unilateral aplasia of the sternocostal head of the *pectoralis major* and an ipsilateral symbrachydactyly with a higher prevalence on the right side.[1] However, Poland anomaly is often associated with other abnormalities, such as hypoplasia or neoplasia of the breast, reduced axillar hair, anomalies of the bony thorax, dextrocardia, renal malformation, neuroblastoma, or leukemia/lymphoma. Most cases of Poland anomaly occur sporadically, but several familial cases have been reported, suggesting a postzygotic mutation of heterozygous individuals. Anomalies of the eye are rare and occur only in combination with Möbius syndrome; these comprise facial nerve palsy with eyelid paralysis or abduction inability.

Case report

We report on a 37-year-old Caucasian male who presented at our clinic with blurred vision in the right eye that had been present for more than 5 years. Medical history revealed that the man suffered from classical right-side Poland anomaly with ipsilateral symbrachydactyly that was surgically reconstructed and ipsilateral renal malformation and malrotation (Figure 1). Visual acuity in the right eye was 20/20, with a defined paracentral scotoma and metamorphopsia in the Amsler chart. The right eye fundus showed a slightly elevated grey lesion in the macular area involving the optic disc and adjacent retina. The lesion comprised a thickened retina with reduced transparency. The retinal vessels in

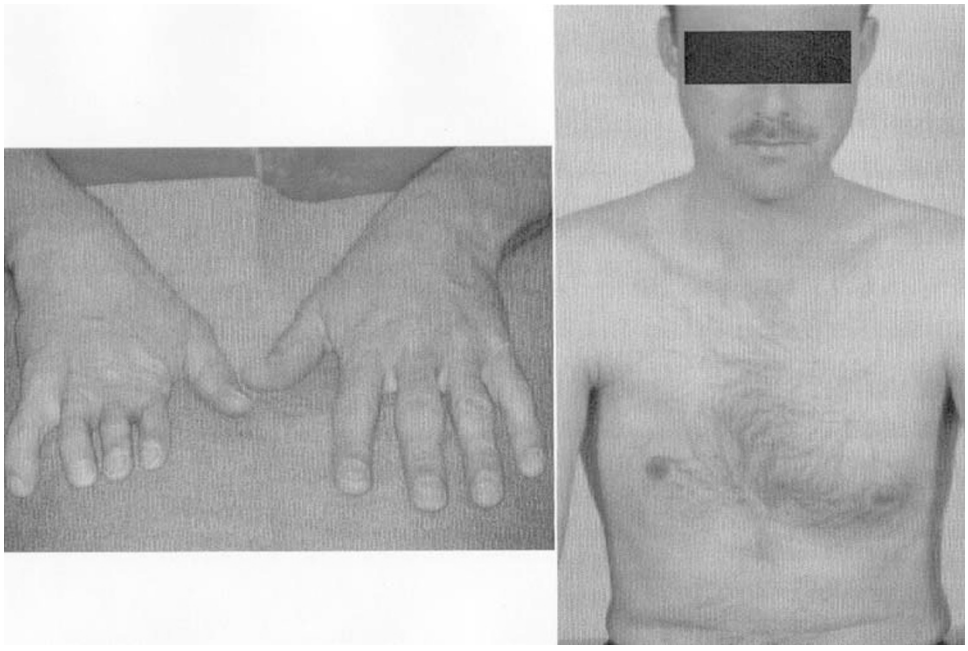

Figure 1 Our patient presenting typical right-sided Poland anomaly with hypotrophy of the *pectoralis major* and partially reconstructed symbrachydactyly.

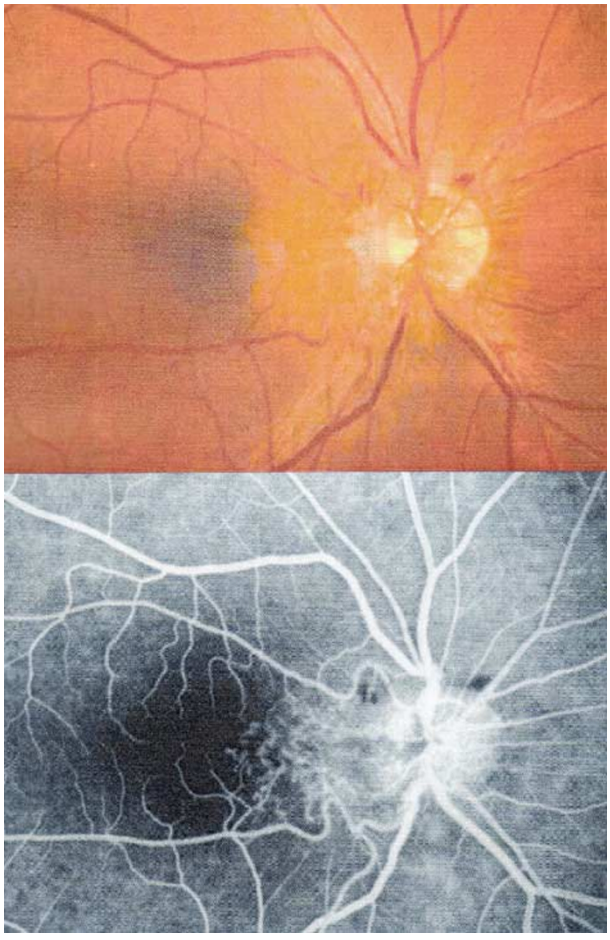

Figure 2 (a) Fundoscopic image showing a grey lesion involving the optic disc and adjacent retina with an epiretinal membrane and wrinkling of the retina. (b) Angiography showing vascular distortion and abnormal capillaries within the lesion.

this area were tortuous and there was an epiretinal membrane with retinal folding (Figure 2a). Fluorescein angiography revealed a network of dilated capillaries in this area with leakage of dye in the late phase (Figure 2b). The left eye was normal.

Comment

In 1973, Gass[2] -first applied the term combined hamartoma of retina and retinal pigment epithelium (CHR-PE) to a series of benign tumours at the level of the sensory retina and retinal pigment epithelium. These tumours consist of thickened semitranslucent retinal tissue, variable pigmentation, abnormal-appearing retinal blood vessels, and some degree of traction at the vitreoretinal interface. In 38% of cases, the CHR-PE is located in the macular area. Characteristic ophthalmoscopic features are slight elevation (80%), vascular tortuosity (93%), and the formation of an epiretinal membrane (78%). However, 13% of all cases of CHR-PE show no pigmentation, as in our case. Fluorescein angiography frequently demonstrates abnormal capillaries with increased calibre, arteriovenous communication, and leakage of dye.[3]

CHR-PE must be distinguished from capillary haemangioma, presenting pure vascular tumours without vitreoretinal interface changes, and from idiopathic epiretinal fibrosis, demonstrating the relatively pure vitreoretinal interface change without vascular changes.

Hamartomas are benign overgrowths of mature cells and tissues that occur normally in the affected area or part. In the case of CHR-PE, one type of tissue (melanocytic, vascular, or glial) often appears to dominate the clinical picture. However, in histopathological examinations Laqua and Wessing did not find a true hamartoma, but rather a disorganization of the retinal architecture with secondary membrane formation and vascular changes, so they suggested classifying these lesions as congenital retinopigment epithelial malformations.[4]

The aetiology of Poland anomaly is unknown. It is assumed that vascular abnormalities at the embryonic stage are responsible for the anomalies: In the 6th week of gestation the pectoral masses split, the interdigital membranes begin to disappear, and vascular differentiation from the six vascular arches to a more mature vascular pattern with distinct vertebral and subclavian arterial branches occurs. According to these events, interruption of the embryonic blood supply may produce various defects depending on the location of the blood-flow restriction.[5]

From the above observations, we suggest that vascular abnormalities observed mostly in the deformed hemithorax in cases of Poland anomaly present a common pathogenetic mechanism, which may also affect ipsilateral ocular development, leading to malformation as shown in the present case.

Only three cases of ocular involvement with Poland anomaly have been reported to date, and these showed ipsilateral coloboma of the iris or of the optic nerve (morning glory syndrome), or ipsilateral juxtafoveal telangiectasis.[6–8] To our knowledge, ours is the first reported case of Poland anomaly associated with CHR-PE on the same side.

There are a few reports on extremely rare cases of CHR-PE associated with neurofibromatosis (NF), mainly promoting NF 2 as the only known possible risk factor for CHR-PE. Furthermore, Alembik and Stoll[9] found one case of a Poland anomaly associated with NF 1. However, our patient had none of the clinical criteria required for a

diagnosis of NF, and also none of his first-degree relatives suffered from NF.

The observations reported here suggest that ocular abnormalities are associated with Poland anomaly. Therefore, careful eye examination including fundoscopy, perimetry, and angiography is recommended in all cases of Poland anomaly.

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