The "Eye" in IP

by

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Incontinentia pigmenti (IP) or Bloch-Sulzberger syndrome is an inherited condition which affects the ectodermal structures (skin, hair, teeth) and also the eyes and nervous system. The condition is inherited as an X-linked dominant disorder which is usually lethal in males (see Landy and Donnai 1993 for review). IP is caused by mutations in the NF-kappaB essential modulator (NEMO) gene. Affected female infants develop characteristic abnormalities of the skin, teeth and hair. However, the ocular and neurological features represent the major cause of concern in IP.

Dr Helen Stewart (Consultant in Clinical Genetics, Oxford) of the Medical Advisory Board has previously summarised the clinical and genetics features of IP which can be seen on this website.

Visual defects can occur in IP and retinal vascular abnormalities of the retinal blood vessels represent the major cause of severe visual impairment. An ocular examination should be performed as soon as possible after birth and IP patients should be followed carefully by an ophthalmologist. Drs. Lewis and Goldberg are both members of the Scientific Advisory Council of the Incontinentia Pigmenti International Foundation and have also posted advice on their website. (scroll to the bottom of the page)

Numerous eye abnormalities in Incontinentia Pigmenti have been reported, but the majority of them result from the abnormalities found in the retinal blood vessels. These retinal vascular abnormalities, which can lead to retinal detachment, represent the major cause of severe visual defects in IP. The mechanisms causing the changes in blood vessels are not fully understood. The natural history of retinal disease in IP is also unknown and optimal treatment of the retinal problems is not clear at present. There are reports of successful treatment of proliferative retinopathy ("overgrowth in the retinal blood vessels") with cryotherapy (freezing treatments) and laser.

A recent study from Sweden of 30 patients with IP has recommended eye screening for all newly diagnosed infants with IP and the female offspring of affected women. The following scheme was suggested: ocular examination as soon as possible after birth, monthly until 3-4 months, three monthly until one year of age, and then bi-annually until 3 years of age. Our knowledge of the natural history of the retinal changes in IP is not complete and the duration of screening remains unclear, therefore ophthalmology review should perhaps be continued on an annual basis throughout childhood. We have recently published a paper on this topic to highlight the importance of an eye assessment to clinicians. [Wong GAE, Willoughby CE, Parslew R, Kaye SB. Importance of Screening for Sight-Threatening Retinopathy in Incontinentia Pigmenti. Paediatric Dermatology 2004; 21(3):242-245.].

An ocular assessment can be arranged by referral to the local ophthalmology department from a GP, clinical geneticist, paediatrician/neonatologist or dermatologist.

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