Incontinentia pigmenti is not common. It is X-linked dominant and is lethal in most affected male infants. Usually the males with the disease are stillborn. There are dermal, central nervous system, ocular, and dental involvements with this disease.

The skin lesions progress through several stages. At birth the skin has blisters that give rise to warty outgrowths. These growths then subside and become hyperpigmented whorls, which may remain for life. Ocular problems are seen in about one-third of patients. Some of the more serious causes of vision loss in these patients include cataracts, retinal neovascularization, chorioretinitis, uveitis, pseudoglioma, retinal detachment, optic atrophy, and a combination of these problems. Strabismus and nystagmus can also occur. Some investigators have reported whorls of pigment in the fundus.

The skin histopathology shows macrophages containing dystrophic cells and melanosome complexes. In the blister stage, eosinophilia may be associated.

Selected Readings

FIGURE 57-1.
Warty outgrowths follow resolution of the blisters at 6 weeks. Here the warts can be seen on the patient’s fingers.

FIGURE 57-2.
Strabismus and a pseudoglioma.

FIGURE 57-3.
Left eye has an opacity behind the lens that is vascular. Ultrasonography showed total detachment.