

SUMMARY

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PXE is a hereditary disease characterised by calcification of elastic fibers in the dermal ocular and vascular tissue.

It was considered as an autosomal recessive trait, (McKusick, 1966) however recently Pope (1975) proposed its genetic heterogeneity. Four types are described:

Dominant Type I: In which there are elastic cutaneous changes, common vascular complications, and severe choroidoretinitis.

Dominant Type II: Cutaneous lesions are atypical, may be absent, no vascular complications and mild retinal changes.

Recessive Type I: It is the most common one, the clinical spectrum comprises the three systems yet in a mild form.

Recessive Type II: Only skin rash.

Muskusick (1966) reported that females are more frequently affected than males.

Calcification of dermal elastic fibers is the earliest detectable change in the dermis. The fibers as well as Bruch membrane are more fragile.

The age of detectability varies from 1st. to the 3rd. decade.

The minimal diagnostic criteria are the angioid streaks and PXE skin changes in the absence of Paget's disease of bones. The skin lesions are usually observed by the second decade at the latest and are most striking around the ~~neck~~ and in the axillae. The skin in the groins, Penis and the periumbilical area may be affected also. The skin has yellowish color due to retiform soft papules. The affected area later becomes lax, redundant and relatively inelastic. Exaggeration of nasolabial folds and chin creases occurs.

In the eyes the hallmark is angioid streaks which are irregular streaking radiating from the disc and lying opposite the retinal vessels. Hemorrhages also occur in the fundus and threaten vision.

Calcification in the elastic fibers of vessels is accompanied by rupture or occlusion giving rise to weak or absent peripheral pulse, mild hypertension, intermitted Claudications, angina pectoris and gastrointestinal hemorrhages (Eddy and Farber, 1962).

As regard treatment, primarily by genetic counseling, vitamin E was recommended without apparent benefit. Surgery may be performed removal of redundant skin.