Acanthocytosis

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Summary

A patient is described with hypo- β -lipoproteinaemia and pen-central tapetoretinal and tapetochoroidal degeneration, in whom fluorescein angiography revealed marked atrophy of the choro-capillaris, which could not be demonstrated in any other way.

The hereditary pattern given in the literature suggests an autosomal recessive gene; in a son and daughter of the patient hypo- β -lipoproteinaemia was found as part of the syndrome.

Discussion

Waardenburg: It seems to me that you have mentioned practically everything that is characteristic of this new syndrome. As the slides you have projected only showed the posterior pole, however, it is difficult to judge whether retinopathia punctata albescens is present. There appear to be quite a number of extremely small, very white dots, which remind me of Bailliar's capillarosis. Mention is made in the literature either of an atypical retinopathia pigmentosa or occasionally of 'retinitis punctata albescens'. It is difficult to say, however, if the latter is not also somewhat atypical. I should therefore like to know if, in your case, the white dots were also present in the periphery. It is also significant that you—in agreement with Salt et al.—found a lesser degree of β -lipoproteinaemia in the two children, as symptom of heterozygotism, than in the homozygotic father. In such families it is of the greatest importance to be able to recognize heterozygotic carriers.

Craandijk replies: The crystals could only be seen centrally. In the periphery there was some migration of pigment but no crystals, i.e. white dots.

Oosterhuis: The brilliant fluorescein photographs are extraordinarily clear and show a very detailed pattern of anomalies. It strikes me that, in the demonstrated slides, the pattern of the anomalies in the fluorescein photographs is very different from that seen in the ordinary black and white photographs and the colour transparencies.