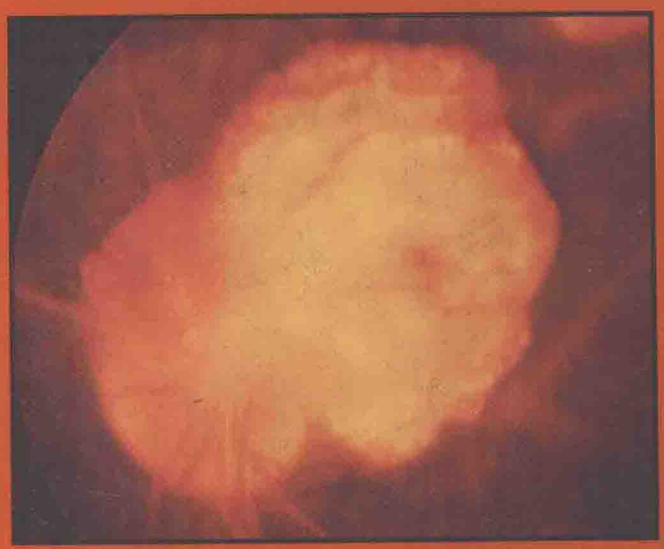
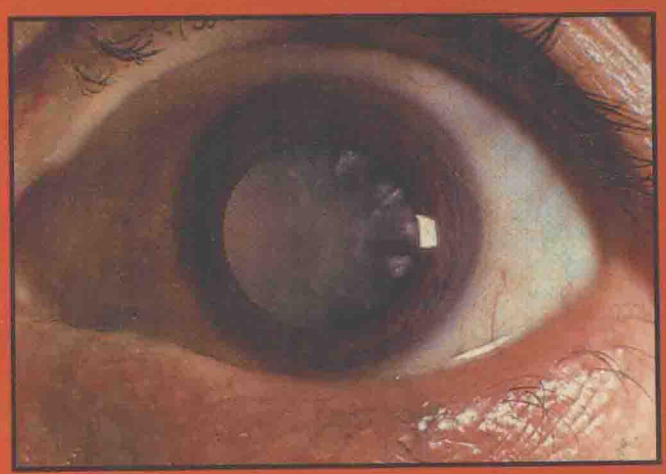
A Colour Atlas of
The Eye and Systemic Disease





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The Eye and Systemic Disease

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To our patients

Introduction

This Atlas aims to illustrate abnormalities of the eye associated with common systemic diseases. In addition it demonstrates some of the 'classic' eye signs of rarer disorders which all physicians are expected to recognise, but seldom have the opportunity to encounter.

Although designed primarily as an aid for those engaged in postgraduate studies in ophthalmology and internal medicine, it also provides a refresher course for clinicians already established in these specialities and in allied disciplines such as rheumatology, dermatology, endocrinology and neurology.

Erna E Kritzinger
Barry E Wright

1: Inborn errors of metabolism

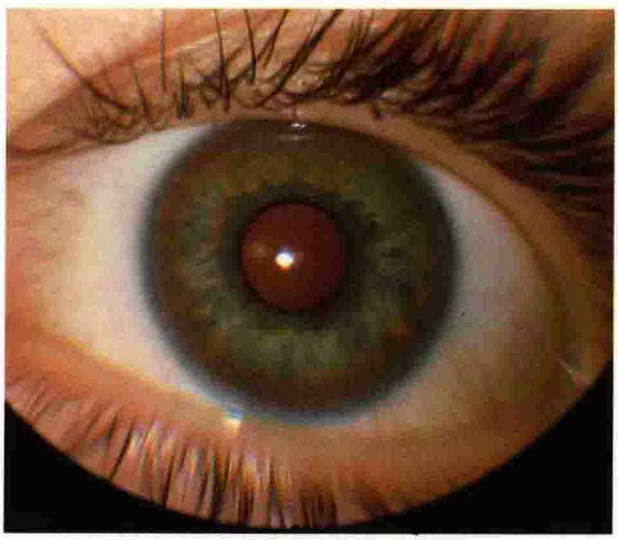
Of the many inherited metabolic disorders with associated ocular signs, those occuring in Wilson's disease are among the few which provide the basis for a specific diagnosis. Abnormal amino acid metabolism is represented by homocystinuria and albinism and abnormal carbohydrate metabolism by galactosaemia. Disorders of lipid and lipoprotein metabolism are among those most frequently presenting with ocular abnormalities and these are illustrated by hyperlipidaemia. The metabolic factors underlying retinitis pigmentosa remain to be elucidated although, in some instances, Vitamin A deficiency is implicated.

Wilson's disease (hepatolenticular degeneration)

In this metabolic disorder, transmitted as an autosomal recessive trait, there is a deficiency of the copper-binding plasma protein ceruloplasmin, resulting in deposition of copper in the brain, liver, kidneys and eyes.

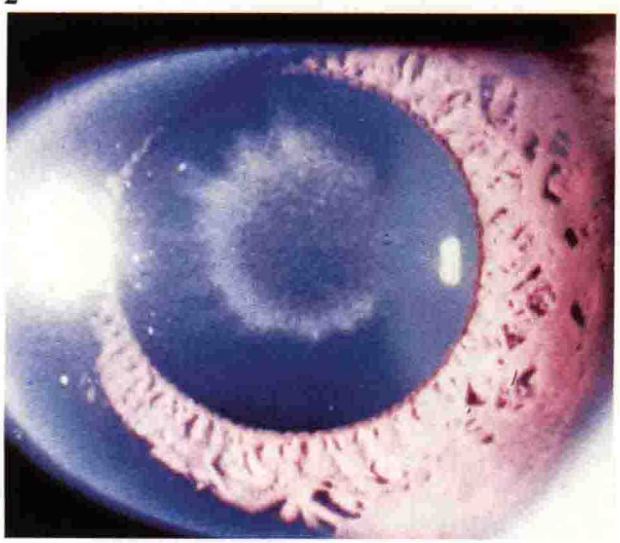
The ocular abnormalities include a Kayser-Fleischer ring in the cornea and 'sunflower' cataract; both are pathognomic of Wilson's disease.

1



1 A Kayser-Fleischer ring. This is brought about by deposits of copper in Descemet's membrane, and is seen as a rusty brown ring at the corneal periphery.

2



2 'Sunflower' cataract. Deposits of copper hydrate form a radiating pattern on the anterior capsule of the lens; normally vision is not seriously affected.

Homocystinuria

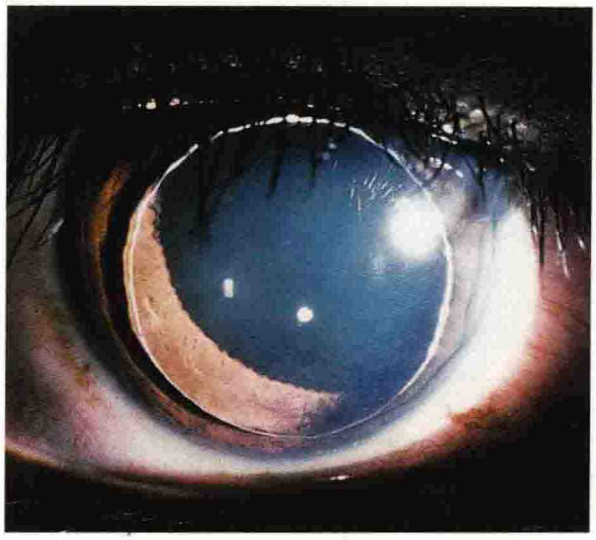
This is a metabolic disorder of amino acid metabolism in which deficiency of the enzyme cystathionine synthetase affects the normal formation of connective tissue and leads to an accumulation of homocystine in the blood and urine and methionine in the blood: it is transmitted as an autosomal recessive trait.

The patients, about 50 per cent of whom are mentally retarded, show skeletal and ocular changes similar to those occurring in Marfan's syndrome (page 15) from which homocystinuria has to be differentiated.

Dislocation of the lens (ectopia lentis) is a common presenting feature of both conditions. In homocystinuria the lens typically dislocates infero-nasally and may enter the anterior chamber of the eye. The dislocated lens predisposes to secondary glaucoma and retinal detachment and frequently becomes cataractous. Other ocular abnormalities include myopia, congenital glaucoma (buphthalmos), peripheral retinal degeneration and optic atrophy.

Thromboembolic disease is an additional factor associated with homocystinuria, and makes ophthalmic surgery hazardous.

3



3 Ectopia lentis in homocystinuria. The lens has dislocated into the anterior chamber of the eye and lies in front of the pupil. (Illustration by courtesy of Miss E M Eagling.)

Albinism

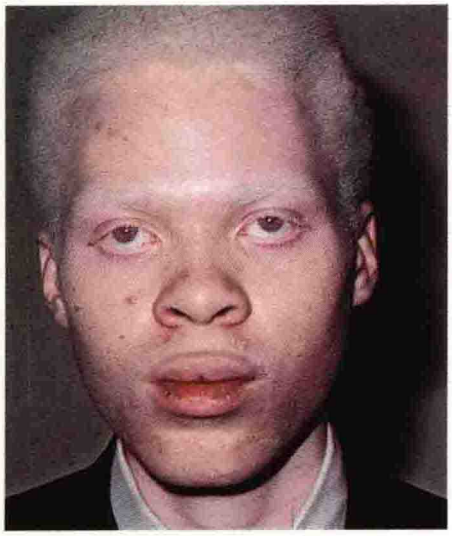
There are three types of albinism, all of them caused by abnormal melanin production.

Oculocutaneous (tyrosine negative) albinism is caused by absence of the enzyme tyrosinase, blocking the conversion of tyrosine to dopa in the biosynthesis of melanin. This is the most extreme form of albinism, individuals being characterised by white hair, pink skin and cutaneous photosensitivity. The irides are blue-grey and diaphanous, the retinal red reflex is prominent and the fundus lacks pigment. Visual acuity is reduced due to photophobia, hypermetropia and a high degree of astigmatism. Nystagmus is common.

Oculocutaneous (tyrosine positive) albinism resembles the tyrosine negative form of the disease in that it is carried as an autosomal recessive trait; individuals have normal levels of tyrosine, however, and are less affected. Skin, hair and ocular pigmentation increase with age. Visual acuity is moderately reduced because of photophobia and nystagmus but tends to improve throughout childhood.

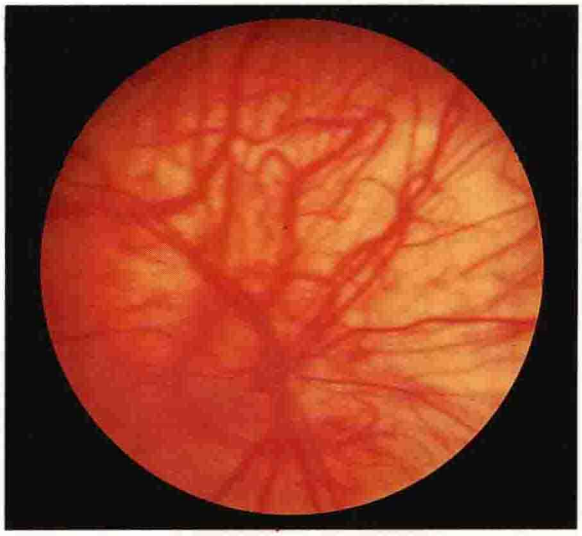
Ocular albinism may be inherited either as an X-linked or autosomal recessive character and affects the eyes only. The clinical signs are similar to those described for tyrosine negative oculocutaneous albinism; although the irides may be pigmented, visual acuity is reduced. Visual acuity is normal in female carriers of the disease, but they have diaphanous irides and pigmentary abnormalities of the retina.

4



4 Oculocutaneous (tyrosine negative) albinism in a negro.

5



5 The fundus in albinism lacks pigment; retinal and choroidal vessels show up against the underlying white sclera.

Galactosaemia

There are two forms of galactosaemia, both of them autosomal recessive inherited abnormalities of carbohydrate metabolism.

In 'classic' galactosaemia, deficiency of the enzyme galactose-1-phosphate-uridyltransferase leads to accumulation of galactose-1-phosphate and galactose in the blood and other tissues: cataract, mental retardation, hepatomegaly, jaundice and malnutrition result.

In the second, milder form of the disorder there is a deficiency of the enzyme galactose kinase which results in accumulation of galactose in the blood and other tissues. Cataract formation is the only clinical sign.

Individuals affected with either of these conditions may have cataracts at birth although, more frequently, they develop them within the first few weeks of life. Dietary elimination of galactose during early life may arrest cataractous changes or even bring about a regression.

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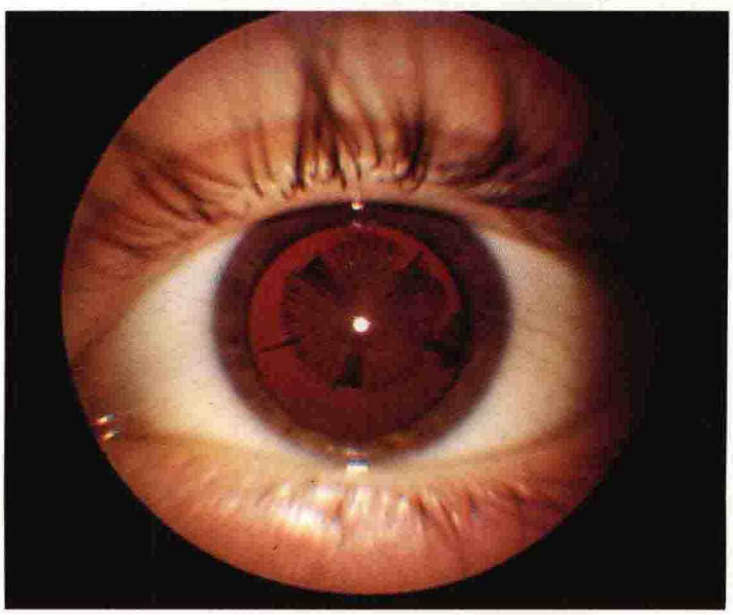

6 Cataract in galactosaemia. The appearance is that of an 'oil drop' in the centre of the lens; zonular opacities may also occur. (Illustration by courtesy of Orthoptic Department, Children's Hospital, Birmingham.)