ICHTHYOSIS VULGARIS ASSOCIATED WITH CATARACT AND NEPHROTIC SYNDROME

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Summary

A 20-year-old female patient of ichthyosis vulgaris with anterior polar cataract is presented. In addition she had nephrotic syndrome. There was family history of ichthyosis but no history of cataract and kidney disease. Relevant literature is reviewed.

Ichthyosis vulgaris is a geno-dermatosis transmitted by autosomal dominant or as recently shown by Wells and Jennings[1] as X-linked recessive trait. Patient with ichthyosis exhibits prominent scales, occuring more or less diffusely all over the body. Usually the scales are large, centrally adherent with loose edges. Apart from being associated with atopy, ichthyosis is often associated with many other genetically determined defects. In 1927, Rud[2] described a syndrome characterised by dwarfism, epilepsy, infantalism, macrocytic anaemia and polyneuritis associated with ichthyosis. Sonneck[3] described a syndrome in which cryptorchidism was associated with ichthyosis. Refsum[4] described a syndrome characterised by retinitis pigmentosa, peripheral neuritis and progressive nerve deafness associated with ichthyosis and now it is known to be defect in phytanic acid metabolism[5]. Sjogren and Larsson[6] described 28 cases with congenital spasticity, low grade mental retardation and ichthyosis. In addition to above mentioned syndromes ichthyosis is also reported to be associated with mental deficiency[7], acrofacial dysostosis and alopecia[9]. Various occular changes like ectropian[10], cataract[11] and some corneal abnormality[12] have been reported with ichthyosis. Cataract associated with ichthyosis vulgaris is a rare occurrence. Uptil now only 9 cases of cataract with ichthyosis have been reported. Herewith we present a case of ichthyosis vulgaris associated with anterior polar cataract. In addition the patient also had nephrotic syndrome. Association of nephrotic syndrome with ichthyosis is not yet reported. However, Goyer et al[8] reported hereditary nephritis, neurosensory hearing loss, prolinuria and ichthyosis.

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Case Report

A 20-year-old single Hindu female was admitted in the Irwin Group of Hospitals, Jamnagar, in February 1972. She had ichthyosis vulgaris since early infancy. The disease used to regress during summer and exacerbate during winter months. She developed generalized anasarca since last one month. Oedema first developed on the face and

gradually spread all over the body. Her parents were normal and had not suffered from any skin disease. One of her brothers out of four siblings also had ichthyosis. Past and personal history was non-contributory.

On examination, patient had mild ichthyosis on extremities (Fig. 1) and back. There was puffiness of face and gross oedema over feet. Blood pressure was 110/80mm of Hg. Pulse was 80 per minute. There was anterior polar cataract in both eyes. Systemic examination did not reveal any abnormality.

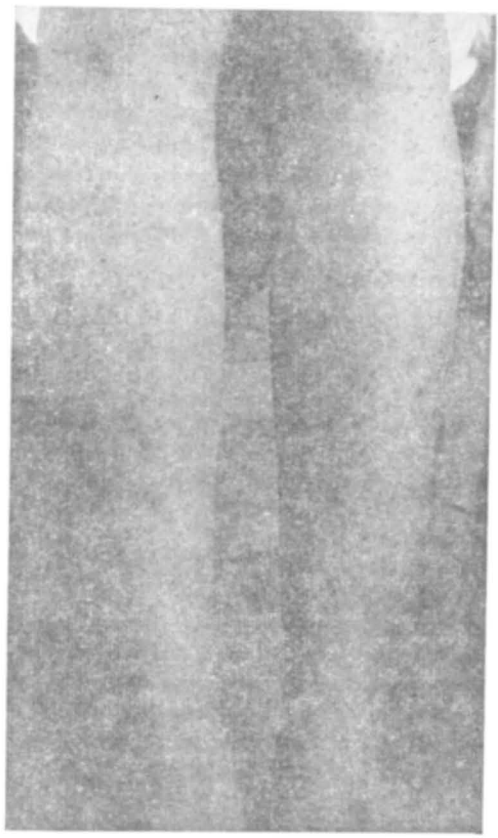

Fig. I

Ichthyosis on the anterior aspect of the legs

Urine examination showed albumin (++++), sugar was absent and microscopic examination revealed granular casts. Haemoglobin was 8.8 G% with normocytic normochromic red blood cells. The total leukocytic count was 4,500/ cmm. with polymorphs 67 per cent, lymphocytes 30 per cent and monocytes 3 per cent. Twenty four hour urinary protein estimation was 9.2 G, E.S.R was 42 mm. at the end of one hour by Wintrob's method. Serum cholesterol was 744 mg%, serum calcium 7 mg%, serum protein 5.2 G% with albumin 2.5 G and globulin 2.7 G%. Blood urea was 20 mg%. Fundus examination did not show any abnormality. Slit lamp examination showed bilateral anterior cuneiform opacities involving cortex anteriorly. The central cortex and subcapsular areas also showed increased density. Skin biopsy obtained from lateral aspect of right leg showed marked hyperkeratosis, absence of granular layer with normal stratum malpighi and flattened rete ridges. Skiagrams of chest and abdomen were normal. Blood examination for L.E. cells was negative. Kidney biopsy could not be performed as the patient did not agree for the same. Patient was treated with diuretics, corticosteroids, high protein diet with salt restriction and haematinics. Patient improved and oedema disappeared after one month of treatment but mild protenuria persisted. Skin condition remained more or less unchanged.

Discussion

Cataract associated with ichthyosis is rare occurrence. Siemens (cited by Jancke[14]) could find only one case of laminated cataract out of 242 cases of ichthyosis vulgaris. Kugelberg (Cited by Jancke[14]) described ichthyosis, "Besnier's prurigo" and cataract in 15 year old patient. There was family history of neurodermatitis, ichthyosis and asthma in several family members. Jancke[14] reported 5 cases of cataract associated with ichthyosis of which 2 were sisters aged 8 and 13 years. Other

3 cases were middle aged, unrelated females. Surprisingly all the 5 were females. Pinkerton[11] reported 2 Japanese boys having ichthyosis with cataract. They were the only children in their families, without any history of ichthyosis or cataract on either the paternal or maternal side. One out of these was 9 year old male child had bilateral lens opacities. The central cortex and subcapsular areas also showed increased density. Other was 12 year old and had bilateral involvement but right was involved more than the left. In both the cases fundus and cornea were normal. Present case was 20 year old female, had ichthyosis since early infancy and cataract was detected on routine examination. In addition she had nephrotic syndrome. There was family history of ichthyosis but no history of cataract and kidney disease in the family. In view of cases reported earlier the relationship of cataract and ichthyosis can be more than fortuitous. However, Siemens[14] believed this to be a mere coincidence and Kugelberg[14] was of the opinion that cataract in his case was probably caused due to associated neurodermatitis rather than ichthyosis. Pinkerton[11] believed that association of ichthyosis and cataract was not because of mere coincidence and suggested thorough clinical and histological examination in cases of cataract, particularly in children to detect mild ichthyosis. We could not come across any report of association of ichthyosis, cataract and nephrotic syndrome in literature. However, association of ichthyosis, heriditary nephritis, prolinuria and neurosensory hearing loss, was reported in family by Goyer et al[13]. Out of 78 cases studied 6 had ichthyosis, of these 2 had all the four manifestations; 2 had ichthyosis and hearing loss while remaining 2 had only ichthyosis. They postulated that these four manifestations were transmitted by incomplete dominant pleomorphic and not completely penetrant trait. The relationship between ichthyosis, cataract and nephrotic syndrome is not known. However, it should be kept in mind that conditions which favour the expression of ichthyosis could also favour the manifestations of other unrelated traits.

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True :

Immunoglobulins of IgA type was the principal or only component in the dermo-epidermal junction of all cases of Dermatitis Herpetiformis, in a series of cases studied by Chorzelski et al. Immunofluroscence was usually microgranular or fibrillary and occurred most intensively in the tips of the papillae. Immunofluroscence was absent or extremely faint in actual vesicular lesions. In cases of bullous pemphigoid Ig G predominated and a continuous line along the basement membrane was seen in all skin specimens. Staining was seen in normal skin as well as the bullous lesions.

Ref: J Invest Derm, 56 : 373, 1971