

Novel Association of Blepharophimosis Syndrome with Xanthoma Tuberosum

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Six year old male referred to our Oculoplasty clinic for bilateral drooping of eyelids was diagnosed to have Blepharophimosis syndrome, (Blepharophimosis, ptosis, epicanthus inversus) (Fig. A) incidentally found to have multiple eruptions over flexure aspect of knees, ankles, buttock (Fig. B, C), arcus juvenilis suggestive of familial hypercholesterolemia. These co-existing autosomal dominant mutations for Blepharophimosis syndrome and Familial hypercholesterolemia in a single patient has never been reported in literature till date.



Fig. 1a: Blepharophimosis syndrome showing bilateral ptosis, Blepharophimosis, epicanthus inversus.



Fig. 2b and c: Xanthoma Tuberosum over flexure aspect of buttock, knees and ankles.

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