

LARGE PLEXIFORM NEUROFIBROMA: UNUSUAL CAUSE FOR BLINDNESS

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ABSTRACT

Plexiform neurofibroma presents anywhere along the course of nerves usually trigeminal nerve. Unilateral amblyopia is caused due to retinal deprivation of sensory sensation. In our case large size of neurofibroma hanging from the supraorbital ridge acted as a curtain for the eye depriving it from sensory stimulation before the age of fixation of response of brain to light, leading to complete blindness.

KEY WORDS: Plexiform neurofibroma, Amblyopia.

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INTRODUCTION

Neurofibromatosis is a genodermatosis of neuroectodermal origin characterized by multiple cutaneous tumours (mollusca fibrosa), pigmented 'cafe au lait' macules, axillary freckles, lisch nodules in iris and variable involvement of central nervous system.¹ This entity may be either asymptomatic or may impair the function, produce disfigurement and be the site of malignant nerve sheath tumours. Rarely may they present with the complication of amblyopia. Amblyopia or 'lazy eye' as it is also called is blurring of vision due to poor transmission

of images to brain. If this occurs early in life before the fixation of sensory areas of brain, it may lead to an irreversible damage to image perception. If this is neglected in early childhood, it may lead to total blindness.

History: Mrs ABC 62 year old female presented to us with a mass hanging from left supraorbital ridge and front and left side of scalp. She had this mass since her birth which gradually increased in due course of time and was large enough to cover her left eye by the age of six years. (Fig-1)

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Fig-1: large neurofibroma as curtain to left eye.

She had difficulty in vision due to obstruction and by age 10 years she had complete loss of vision in her left eye. Her management started with complete history which revealed similar lesion in her younger sister. Clinical examination showed small neurofibromas over face arms etc. Apart from routine investigations CT scan of skull was done where no intracranial extension was found. She was managed by complete excision of the neurofibroma with reconstruction by rotational flaps.

DISCUSSION

Plexiform fibrohistiocytic tumour was first described by Enzinger and Zhang¹⁻³ in 1988 and is characterized by fibrohistiocytic cytomorphology and a multinodular growth pattern. It is due to excessive growth of the neural tissue in the subcutaneous fat. It is commonly seen in connection with the branches of trigeminal nerve.⁴ It is considered to be a hamartoma than a typical tumor. Grossly, the tumour is usually described as being grey-white and firm and ranging in size from 0.5 to 8.0cm. Plexiform neurofibroma presents as a diffuse and elongated swelling along the course of facial nerve trunk/plexus, these tend to infiltrate into deeper structures like fascia, muscles and bone. The genetic defect is localised to chromosome 17 and is transmitted in an autosomal dominant pattern.⁵

Amblyopia is defined as dimness of vision without any pathology in the eye. The problem is caused by either no transmission or poor transmission of the visual image to the brain for a sustained period of dysfunction or disuse during early childhood usually occurring before the age of 8 years of life with first three years being most crucial ones.⁶ The condition will only arise at this young age because most of the visual system's development in humans is complete and "locked in" by 8 to 10 years of age.

Three critical periods of human visual acuity development have been determined. During these time periods, vision can be affected by the various mechanisms to cause or reverse amblyopia. These periods are as follows:

- * Development of visual acuity from the 20/200 range to 20/20, which occurs from birth to age 3-5 years.
- * Period of highest risk of deprivation amblyopia, from a few months to 7 or 8 years.
- * The period during which recovery from amblyopia can be obtained, from the time of deprivation up to teenage or even, sometimes, adult years. It is caused by Anisometropia, Strabismus and Visual deprivation.

Management of amblyopia should start as early as possible and usually it is the cause which has to be treated. The treatment is then directed towards restoring as much vision as possible by forcing the use of the amblyopic eye by occlusion therapy. Occlusion therapy has been the mainstay of treatment. The patient suffered massive myocardial infarction during reversal and we lost her. The aim of this manuscript is to highlight the unusual cause of amblyopia and the fact that early identification could prevent this disabling disease.

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