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Case report: Neurofibromatosis type i

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Abstract
Neurofibromatosis also as Von Recklinghausen was able an association between the cutaneous and tumor lesions. This disorder is divided into neurofibromatosis type 1, neurofibromatosis type 2 and schwannomatosis. Neurofibromatosis type I, in which the nerve tissue grows tumors that may be benign and may cause serious damage by compressing nerves and other tissues. More than half of the people affected by neurofibromatosis have a family history of it with some type of genetic mutation. This paper presents a case of Von Recklinghausen with clinical signs especially in eye. Some treatments for the patient's quality of life are discussed.

Keywords: Neurofibromatosis, cutaneous and tumor lesions, blindness, quality of life.

1. Introduction

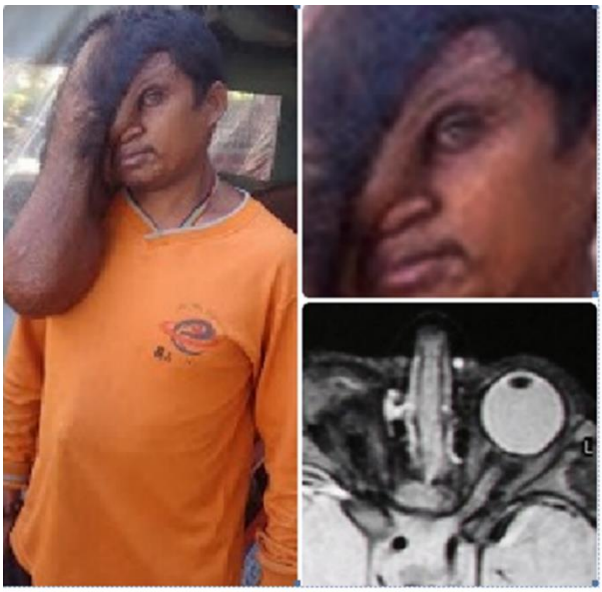
Neurofibromatosis was firstly described by Friedrich Daniel Von Recklinghausen, a German researcher, in 1882 (Spanish Association of Neurofibromatosis, 2001). With 2018 ICD-10-CM Diagnosis Code of Von Recklinghausen disease (neurofibromatosis) is Q85.01 Neurofibromatosis also as Von Recklinghausen was able an association between the cutaneous and tumor lesions (Woodrow, Clarke and Amirfeyz, 2015). This disorder is divided into neurofibromatosis type 1(NF1), neurofibromatosis type 2 (NF2) and schwannomatosis. [1]. More than half of the people affected by neurofibromatosis have a family history of it with some type of genetic mutation. Neurofibromatosis is an autosomal dominant disorder. If only one parent has neurofibromatosis, his or her children have a 50% chance of developing the condition as well. The affected child could have mild NF1 even though inherited from a parent with a severe form of the disorder [1, 2]. Neurofibromatosis type I, in which the nerve tissue grows tumors that may be benign and may cause serious damage by compressing nerves and other tissues [3]. Pathophysiology: The pathophysiology of neurofibromatosis type 1 consists of the NF1 gene protein [4]. This protein is a tumor suppressor, a signal regulator of cell proliferation and differentiation. Schwann cells in neurofibromas have a mutation in the NF1 alleles [5]

Epidemiology NF1 occurs in 1 in 3000 individuals and is equally prevalent among men and women. Furthermore, it is among the most common inherited nervous system disorders [6]. Such individuals have a 10 to 15 year reduction in life expectancy compared to the average person [7].

This paper presents a case of Von Recklinghausen with clinical signs especially in eye and some treatments for the patient's quality of life are discussed.

2. Case Report

A 37 year old man with NF 1 was referring to ophthalmologist for checking low vision progressively during one month previously with mild pain in left eye. General state: Height= 1, 55 meter; Weight=52 kg; Pulse = 78/mn; Arterial tension=125/75 mmHg. Mental = Nothing abnormal detecting (N/A). Ophthalmology examination: This patient with tumor on the right hand side from the frontal head skin. Size tumor has a tube shape. The root- tube is on the head skin with 18mm in diameter and the end- tube is 25mm in diameter. The length of tube is 60cm. The volume of this tumor is about 6-8kg. Right eye (RE) = no ocular. Left eye (LE) =Corneal opacity at 4-7 o'clock. Anterior chamber =clear with slit lamp checking. Reflex oculomotor pupil with light is (+/-.) Press on eye ball= A mild pain (+). Diagnosis: Neurofibromatosis 1-RE= Anophthalmia. Visual acuity (VA) = 0. LE= Uveitis. VA= count fingers at 3 meter. Treatment LE= Steroid included: drop, local suspension 0,5 ml periocular injection one time for 3 days.in the 1st week and 2 times for 2 consecutive weeks. Prednisolone 5ng Per Oral 40mg was used for one week after that drop dose for 2 weeks before stopping.VA of LE is increased to count fingers at 5 meter after 3 weeks treatment. One month later he is not any complain on his ocular problems.



Neurofibromatosis type I

Fig 1

3. 'Discussion

Signs and symptoms of NF1: In early life this disease may cause learning and behavior problems – about 60% of children who have NF1 have a mild form of difficulty in school. In terms of signs the individual might have are the following. Six or more light brown dermatological spots; At least two neurofibromas; At least two growths on the eye's iris; Abnormal growth of the spine (scoliosis) [8].

Ocular problems

- Tumors of the optic nerve (gliomas) occur in about 15% of children with NF1.
- They are often asymptomatic but, over time, tumors may cause visual acuity loss, abnormal color vision, visual field loss, squint, pupillary abnormalities, pale optic disc, proptosis and hypothalamic dysfunction. Risk is highest in those aged under 7 years. Young children rarely complain of early visual impairment and sometimes it is not picked up until it is advanced, with bilateral visual loss. The most common presentation is asymmetrical visual field defects. Optic nerve gliomas occasionally start to cause symptoms in older children or even adults. They can also undergo spontaneous regression.
- Lisch nodules are usually only seen by slit lamp. Occasionally, they can be visible via the ophthalmoscope.
- Patchy choroidal abnormalities retinal vessels are sometimes seen in patients with NF1.
- In NF2, posterior subcapsular or juvenile cataracts can precede CNS symptoms. These cataracts may progress over time, impairing visual acuity. Some have retinal hamartomas or epiretinal membranes that are not always significant to vision
- Diagnosis: CT scan, Radiograph, MRI or CT scan EEG, Slit-lamp examination, Genetic testing, Histology are used in detecting for diagnosis.

Treatment: Surgical removal of tumors is an indication option. The risks involved should be firstly assessed. With regard to optic pathway gliomas, the preferred treatment is chemotherapy. However, radiotherapy isn't recommended in children who present with this disorder. It is recommended that children diagnosed with NF1 at an early age have an examination each year, which allows any potential growths or changes related to the disorder to be monitored [9].

Prognosis In some cases of NF1 can be severely debilitating and may cause cosmetic and psychological problems. In most cases, symptoms of NF1 are mild, and individuals live normal as well as productive lives.

In this case, a 37 year- old- man, there are no Lisch nodules, no glioma, no patchy choroidal abnormalities retinal vessels as well as no cataract by ophthalmoscope and slit lamp. Ocular problems in this case included anophthalmia and uvetits. Anophthalmia is an extremely rare disease and is mostly rooted in genetic abnormalities. It can also be associated with other syndromes. Anophthalmia has been reported to be present in 3 out of every 100,000 births [10].

This case should be done for tumor removing. After tumor removing esthetic surgery -artificial eye for right eye is necessary. Treatment left eye with uveitis by steroid is performed for restoration patient's vision as well. Option treatment will be increased the patient's quality of life.

4, Conclusion

This case of Von Recklinghausen with clinical signs contributed diversifying especially in eye problems. Some treatments are discussed by increasing visual acuity may be helping this patient's quality of life despite of blindness in both eyes.

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