ORIGINAL ARTICLE

Von Recklinghausen Syndrome with Right-sided Basal Ganglion Abnormalities on Brain Magnetic Resonance Imaging

Aamir Jalal Al Mosawi[1,2]

[1]Department of Pediatrics and Pediatric Psychiatry, Children Teaching Hospital of Baghdad Medical City, Bab Al-Muadham Baghdad, Iraq, [2]Iraq Headquarter of Copernicus Scientists International Panel, Baghdad, Iraq

ABSTRACT

Von Recklinghausen syndrome is a complex genetic disorder that ranges from mild hardly noticed condition to an extremely terrifying progressive disorder with numerous complications and serious disfigurement of the body caused by generalized or huge growth of tumors. The condition was named after Friedrich Daniel Von Recklinghausen who published in 1882 a detailed description of the disorder including information from the previous literature. However, the condition has not been well described or documented in Iraqi children despite it is believed that the condition was observed.The main aim of this paper is to describe a unique case of Von Recklinghausen syndrome in an Iraqi child associated with the right-sided basal ganglion abnormalities on brain magnetic resonance imaging.

Key words: Basal ganglion abnormalities, brain magnetic resonance imaging, childhood, Von Recklinghausen syndrome

INTRODUCTION

Von Recklinghausen syndrome is a complex genetic disorder that ranges from mild hardly noticed condition to an extremely terrifying progressive disorder with numerous complications and serious disfigurement of the body caused by generalized or huge growth of tumors. The condition was named after Friedrich Daniel Von Recklinghausen who published in 1882 a detailed description of the disorder including information from the previous literature. However, the condition has not been well described or documented in Iraqi children despite it is thought that the condition was observed.[1-3] The aim of this paper is to describe a unique case of Von Recklinghausen syndrome in an Iraqi child associated with the right-sided basal ganglion abnormalities on brain magnetic resonance imaging (MRI).

The syndrome is also called type-1 Neurofibromatosis, generalized neurofibromatosis, and multiple neurofibromatosis. It is associated with light brown spots on the skin. Nerve tissue tumors (neurofibromas) that are generally benign, but can cause serious damage by compressing nerves and other tissues. It is also associated with neurofibromas which are tumors involving supporting cells in the nervous system rather than the neurons. In addition, scoliosis, and freckles in the axilla and groin are also observed in this syndrome.[1,4,5] The symptoms in this type are often present at birth or develop before the age of ten years.

The condition characteristically worsens with time, but most patients with this type have a normal life expectancy.

MATERIALS AND METHODS

H.J.M was born in 2005 and was first seen during October 2018 at the age of 13 years due to deterioration in school performance. He was still studying at the fourth-grade primary school despite he should have finished the sixth-grade

Address for correspondence:
Aamir Jalal Al Mosawi, Department of Pediatrics and Pediatric Psychiatry, Children Teaching Hospital of Baghdad Medical City, Al-Muadham Baghdad, Iraq. E-mail: almosawiAJ@yahoo.com

primary school.

The boy had soft tissue mass on the right forehead [Figure 1], and multiple café au lait spots over many parts of his body including back of neck, back, anterior chest wall, abdomen, arms, and upper thigh [Figure 2].

The parents were relatives and the boy had four healthy siblings but was not that good at school including:

- A sister who was born in 2001 was still studying in the third-grade intermediate school (she should have already finished the fifth-grade secondary school)
- A sister who was born in 2002 was still studying in the second-grade intermediate school (she should have already finished the fourth-grade secondary school)
- A brother who was born in 2004 was studying in the third-grade intermediate school
- A brother who was born in 2008 was studying in the third-grade primary school
- Ophthalmological examination includes fundoscopy which showed normal findings with no evidence of Lisch in the iris.

RESULTS

Nothing significant was found on abdominal ultrasound.

Ultrasound of the soft tissue at the forehead showed localized thickened skin layers with increased subcutaneous tissues in an area of about 2.5 cm × 3 cm.

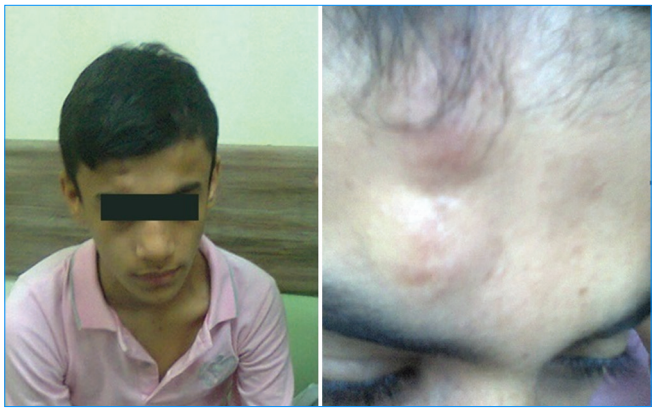

Figure 1: The boy had soft tissue mass on the right forehead

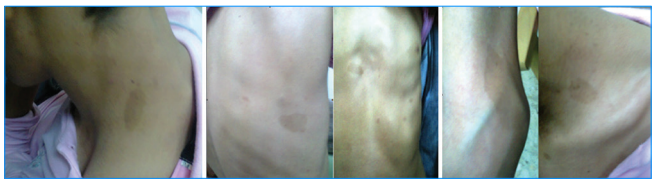

Figure 2: The boy had multiple café au lait spots over many parts of his body including the back of neck, anterior chest wall, back, arms, and the supraclavicular region

Brain MRI showed normal cerebellum and brain stem, and symmetrical appearance of the ventricular system.The optic nerve and chiasma were both normal. Most importantly, the right sided basal ganglion was hyperintense on T2W suggesting demyelination.

DISCUSSION

The three main types of neurofibromatosis include:[1-5]

Type-1: Neurofibromatosis which is also called Von Recklinghausen syndrome, generalized neurofibromatosis, and multiple neurofibromatosis. It is associated with as follows:

Light brown spots on the skin, nerve tissue tumors (neurofibromas) that are generally benign but can cause serious damage by compressing nerves and other tissues, neurofibromas are tumors involving supporting cells in the nervous system rather than the neurons, scoliosis, and freckles in the axilla and groin the symptoms in this type are often present at birth or develop before the age of 10 years.

The condition characteristically worsens with time, but most patients with this type have a normal life expectancy.

Type-2: Neurofibromatosis is associated with as follows: Bilateral benign acoustic neuromas which are tumors of the vestibulocochlear nerve or the eighth cranial nerve, hearing loss, cataract balance problems, flesh-colored skin flaps, and muscle wasting.

The symptoms in this type may not become apparent before early adulthood, and the condition has a variable course, but it may increase the risk of early death as it may cause damage to nearby vital structures such as cranial nerves and the brain stem.

The third type is schwannomatosis associated with painful schwannomas of the spinal and peripheral nerves. Localized types of neurofibromatosis without the light brown spots on the skin characteristic of type 1 neurofibromatosis have also been reported.

CONCLUSION

The patient reported in this paper represents the first case of childhood Von Recklinghausen syndrome associated with the right-sided basal ganglion abnormalities on brain MRI.

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