

Report Case of Van Der Knaap Syndrome in Two Ecuadorian Siblings

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Abstract

Van Der Knaap syndrome is a very uncommon disease seen mainly in the Aggarwal community in northern India. The characteristics of this disease are early onset macrocephaly with mild motor developmental delay, progressive gait alterations, muscular stiffness (spasticity), progressive ataxia, sporadic seizures and usually mild cognitive impairment (MCI) which is of late appearance. Only a few cases of this pathology have been reported in worldwide literature. For the knowledge of all, there are no reports so far of two siblings of parents in which there is apparently no Indian descent or consanguinity. In this case the brothers started with abnormal growth of the cephalic perimeter, difficulty to start the march which progressively got worse, muscular stiffness was developed while intellectual functioning was preserved for several years after onset of the disorder. Was more notorious in one of the two brothers in terms of motor and speech deficit.

Keywords: Macrocephaly; Megalencephalic leukoencephalopathy; Subcortical cysts; Leukodystrophy; MLC1

Introduction

Van Der Knaap syndrome is a neurodegenerative rare disease, it is an autosomal recessive syndrome and its gene is in the chromosome 22 qtel [1]. MLC has a very low incidence around the world, in which very few cases are reported. MLC is characterized by the altered cephalic development (macrocephalic) at moment of birth or it can appear during childhood. Like certain chromosomal pathologies, it is also characterized by presenting in ethnic groups in which consanguineous family exists, like the city of Agrawal in northern India in which the syndrome was reported for the first time [2].

This rare disease is usually characterized by findings in the cognitive development of the affected individual, mild delay in motor functions and the presence of seizures [3]. Cerebral alterations such as subcortical cysts can affect group of muscles and the loss of acquired movements. The first signs which appears more frequently is muscle stiffness and will obviously reflect on mobility, certain patients progressively lose the ability to wander or move from a very early age, which often needs wheelchair support, but others manifest these motor disorders in adulthood. They may also develop dystonia, athetosis, swallowing problems and dysarthria. These patients despite having defects in brain mass only show mild cognitive impairment referred by Batla, Pandey and Nehru [4], the slightest head trauma can lead to or trigger motor alterations and some of them also can trigger seizures.

In certain reported cases, the magnetic resonance findings present the bulging white matter and certain symmetrical changes above the tentorium. The white substance may or may not be diffusely arranged in the cerebral hemispheres with a relative preservation of the central structures of the white matter such as the corpus callosum, the internal capsule and the brainstem [5]. The presence of subcortical cysts is usually found in several regions, most frequently the frontoparietal region. Clearly we can observe the cerebral atrophy and the presence of cysts of different sizes which as they grow can develop more symptomatology.

Case Report

A 13-year-old boy, the oldest of two siblings, they were born of parents in which there is no consanguinity, in Guayaquil, Ecuador which showed increase in their cephalic perimeters since their childhood, at the age of 5 months he had no other features but the enlargement of the head with 51.0 cm of circumference. After two months, 3 cm more grew of his head circumference and so on was gradually growing. He was sent

to do an MRI and the results indicated there was an alteration of the white matter with cysts. There was no alteration in the blood laboratory results.

He was referred to a neurologist, which gave him the diagnosis of Alexander syndrome which is described in the literature as a differential diagnosis. At the age of 8 months he was referred to Miami's children hospital in which was diagnosed of Van der Knaap syndrome. Mildly delayed mental and motor milestones, and progressive ataxia since he was 5-years-old. There is history of seizures in the past that developed after a traumatic head injury after falling from his own height at the age of 8. Physical examination we were able to observe that the vital signs are within the parameters. Size and weight were in percentiles suitable for their age. The cephalic diameter was 61 cm (above the 95th percentile).

Neurological examination showed mild cognitive impairment (MCI). There was no deficit or alteration of the cranial nerves, scale of Daniels 4/5 in muscular strength in the four extremities. Marked dysarthria was present, the tendon reflexes were energetic, both, deep and superficial reflexes, and extensor plantar responses were also present. His gait was ataxic which ultimately forces him to walk with support (wheelchair). His tutors refer he has seizures only with he hits his head.

The magnetic resonance of the brain in Figures 1-3 performed in both the coronal, axial and sagittal sections showed alterations in the gray and white matter, evidencing bilateral subcortical cysts in the frontoparietal region.

The genetic analysis of the pathology or its follow-up was not carried out due to the limited availability since the family could not pay for the follow-up, for which reason it was only explained that the diagnosis was based on clinical data, traits and results of the nuclear magnetic resonance. The cabinet exams included complete blood count, urinalysis and metabolic profile, which showed no abnormalities, EEG

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Received February 19, 2018; Accepted May 16, 2018; Published May 28, 2018

Citation: Moncayo JA (2018) Report Case of Van Der Knaap Syndrome in Two Ecuadorian Siblings. J Clin Case Rep 8: 1122. doi: 10.4172/2165-7920.10001122

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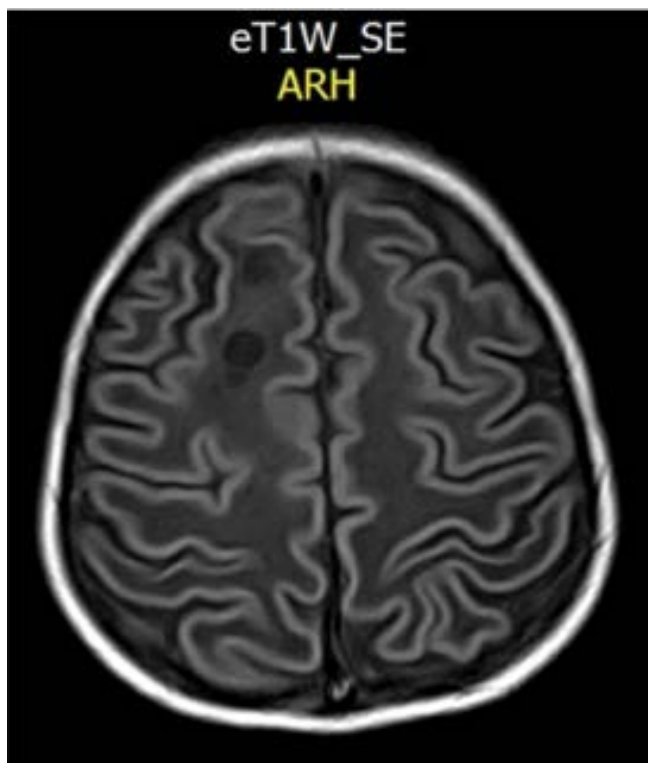


Figure 1: The magnetic resonance of the brain that performed in both the coronal, axial and sagittal sections.



Figure 3: The magnetic resonance of the brain that showed alterations in the gray and white matter, evidencing bilateral subcortical cysts in the frontoparietal region.

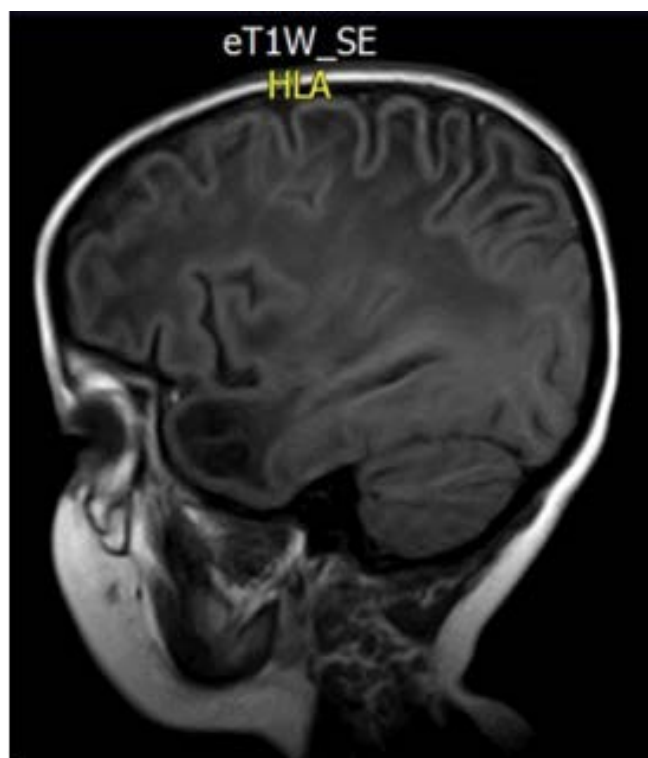


Figure 2: The magnetic resonance of the brain.

was never performed because, according to the literature, seizures are due to cysts in the subcortical area, so the doctors decided not to do it since it was not in the protocol, only seizures should be treated but is not necessary to perform an EEG.

Parents and immediate family members were told what palliative care would be like and how to avoid complications. Something very important that was indicated to them was the control of head trauma and medicine for seizures as well as digestive enzymes to control the intestinal transit since they referred occasional constipation, Currently, the patient is confined to a walker, language therapy and physiotherapy.

Discussion and Conclusion

The Van Der Knaap disease or syndrome is the most prevalent leukodystrophy with megalencephaly observed in Asian descendants. In my study, the oldest of the two siblings belonged to the same family and was not the product of parents with consanguinity or Asian descent. For any individual that presents notable signs or symptoms including images there must be an early diagnosis of the pathology and never rule it out in order to initiate an early and timely treatment. which with this we can decrease the Impaired neurological function and early rehabilitation can prolong outpatient life. However, in the present case, the traumatic incident seems to have caused the outcome and progression of the disease as well as wandering trawling.

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