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VAN DER KNAAP DISEASE PRESENTING WITH ATYPICAL FEATURES AND RADIOLOGICAL FINDINGS: RARE CASE REPORT

Medicine

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ABSTRACT

A 21 yrs old female born out of non-consanguineous marriage presented with progressive neurological symptoms like chronic ataxia with bilateral lower limb weakness. Patient had history of similar complaints since last 4 years. During childhood parents mentioned about history of delayed milestones, pes cavus , superficial hemangioma during early years of life but without history of seizures. Patient underwent MRI Brain with contrast with findings suggestive of Megalencephalic leukoencephalopathy with subcortical cysts (Van Der Knaap disease). Classically this disease presents at birth and during infancy in children born out of consanguineous marriage with seizures are recurrent features. Thus this case highlights atypical features of Van der Knaap disease[2].

KEYWORDS

INTRODUCTION:
Van der knaap disease is a rare autosomal recessive disorder seen in children born out of consanguineous marriage[1]. It generally presents at birth or early childhood likely infancy. Patient presents with megacephaly with progressive increase in size along with delayed milestones and recurrent seizures[3]. Above stated features are due to leukodystrophy with subcortical cysts labelling it as Megalencephalic leukoencephalopathy with subcortical cysts. Some patients can present with delayed onset neurological worsening with spasticity and epilepsy. Till this date no cure has been found for the disease with supportive care and symptomatic management plays crucial role in treatment.

Case Presentation:
A 21 years old female born out of non-consanguineous marriage presented with history of progressive neurological symptoms as chronic ataxia, difficulty in walking and stiffening of lower limbs. Patient had history of similar complaints since she is 4 years of age. Patient had h/o delayed milestones, h/o bilateral lower limb deformity (pes cavus), h/o superficial hemangioma, difficulty in sitting and standing on own, intellectual disability since birth which later increased in intensity. Patient never had history of seizures in childhood or at current episode.

General Examination-
On examination patient was conscious and oriented to time place person
Pulse 80bpm Bp 116/80mmhg
Spo2 98% on room air
Hemangioma on rt side of neck and right arm.
Café au laits spots on left wrist dorsum of hand.
Systemic examination

Central Nervous System-
1. Higher mental functions- Normal
MMSE - 28/30

2. Hypertonia in both lower limbs Rt> Lt
3. Power

	Rt	Lt
Upper limb	5/5	5/5
Lower limb	3/5	4/5

4. **Plantar Reflex-** extensor
5. Pes cavus positive
6. **Sensory System -** Normal on both sides
7. **Cerebellum-** No signs of cerebellar involvement and Gait was also normal
8. **Cranial Nerve-** No signs of cranial nerve involvement
9. No signs of involvement of extrapyramidal system
10. No signs of involvement of Autonomic nervous system

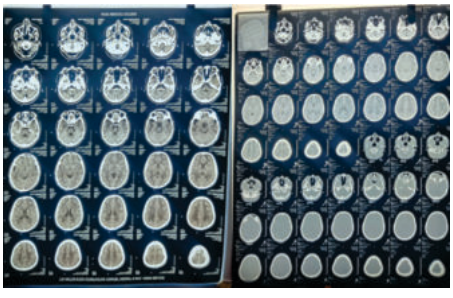
CVS - Heart sounds normal, no murmur
RS- Breath sound bilaterally equal No adventitious sounds
P/A- Soft Non tender No organomegaly.

Investigations-
Lab Values As Follows
Hb-14.4gm/dl. Leucocyte count - 8400/mcgL

Platelets- 334000/mcgL

AST-32 U/L. ALT-42U/L. ALP- 36U/L.
Creatinine- 0.7mg/dl. Urea- 26 mg/dl

CT Brain Plain
s/o global white matter edema, with accentuated grey white matter differentiation s/o white matter disease



MRI Scan Of Brain With Screening Of Whole Spine -
T1, T2 Dwi, Flair, Gre Sequences In Multiple Planes On 1.5t Machine. Bilaterally symmetrical altered signal intensity changes noted involving subcortical U fibers in bilateral cerebral hemispheres appearing hypointense on T1, hyperintense on T2 and FLAIR with restricted diffusion on DWI with high ADC values (T2 shine through). No e/o of blooming on GRE images.

Bilateral subcortical cysts noted in anterior temporal lobes showing CSF signal intensity on all sequences.

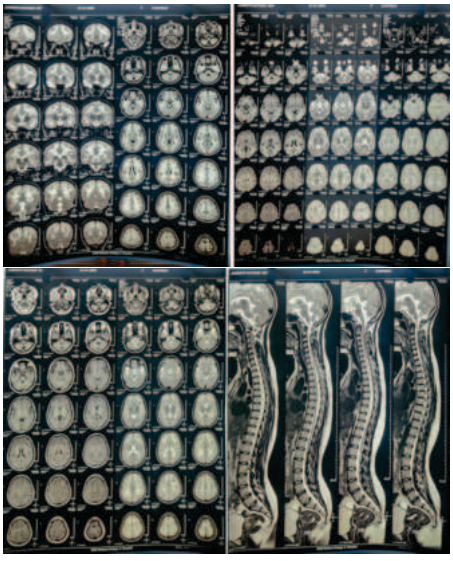
There is sparing of corpus callosum, putamen, caudate nucleus and internal capsule in both cerebral hemispheres.

No e/o involvement of periventricular deep white matter. F/s/o Megalencephalic leukoencephalopathy with subcortical cysts (Van Der Knaap disease)[4].

These features to be differentiated from adult onset leukodystrophies such as cystic leukoencephalopathy without megalencephaly and

metachromatic leukodystrophy. The brain stem appears normal. There is no shift of the midline structures. Posterior fossa structures are normal.

No evidence of intra or extra axial hemorrhage Dural venous sinuses show normal flow void signal. Screening of whole spine: No significant abnormality.



DISCUSSION:

Van der knaap disease is a rare clinical scenario which presents at birth and in some cases might present as progressive neurological symptoms at later ages. It is found more in consanguineous marriages and seen in some communities like Agarwal community in India[5]. Seizures is one of the prominent feature in this disease contributing to more than half of total cases; but as this case is considered seizure was not a feature neither in childhood nor in younger age while chronic ataxia and spastic paraparesis were predominant features. This disease to be differentiated from adult onset leukodystrophies. Thus this case highlights atypical features of van der knaap disease and makes it one of the rare case.

CONCLUSION:

Van der knaap disease is rare disease which usually presents in childhood with seizure and developmental delay can present with atypical features like chronic ataxia and spastic paraparesis without seizures in adults and to be differentiated from adult onset leukodystrophies.

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