

Patient Name: Silla Khaled Mohamed Al Sayed Ateya	Patient ID: 787432
Ref. Phy.: Sollaf Mohamed Al Sayed	Study Date: 06/05/2024

(DOB: Oct 14, 2012)

Medical Report

Dear Prof. Dr. Solaf Mohamed, thank for referring this case of 11 years old female coming with diagnosis of Biotenidase deficiency.

- **The first MR study at outside center of 25-10-2022** is somewhat suboptimal and with not all the brain images provided, however, there is a definite focal zone of signal alteration at the tectum of midbrain together with its swelling best demonstrated on the sagittal FLAIR sequence and marked by dark green tag and there is tendency for diffusion restriction at these lesions denoting activity.
- No other brain parenchyma focal lesions and this with or without contrast to account for contrast behavior.
- No MR evidence of brain atrophy.
- No MR evidence of delayed white matter myelination.
- Normal MR appearance of the sella turcica and craniocervical region.
- As regards the spinal cord, there is a long segment of affection from C1 through C7 best on the FS T1 WIs being of faint high signal and associated with mild cord swelling (orange tags).
- Otherwise, the dorsal cord, conus medullaris and cauda equina are of normal MR appearance.
- No bone marrow lesions.
- No disc bulgings seen.
- **The subsequent MR studies of 1-2024** at our center showing regressive pattern regarding the brainstem mainly tectal plate lesion.
- There is just barely seen faint high signal alteration at the periaqueductal region and just anterior to it on the FLAIR sequence and this is marked by light green tag as representative lesions seen elsewhere in the brain and again no MR evidence of brain atrophy or of delayed myelination.
- Another similar lesion is seen at the lower aspect of the medulla and better demonstrated actually in the sagittal images of the cervical spine also marked by green tags.

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- The MR of the spinal cord and in comparison to the prior study showed the following:
 - Mostly regression of the cervical cord swelling and of the high signal alteration at the cervical cord with noted linear path along the lower aspect of the medulla and upper most aspect of cervical cord marked by green tags.
 - On the other hand de-novo occurrence of similar faint high signal patch within the cord at D3 level marked by red tags on the sagittal T2 WIs (this lesion was not properly identified in the study of 2022 partly due to difference in machine and technique).
 - Normal MR appearance of other parts of cord, conus medullaris and cauda equina.
 - No bone marrow lesion.
 - No disc lesions but mildly exaggerated normal dorsal lordosis and mild rightward convex dorsal scoliosis seen.

OPINION:-

- The finding in the Biotenidase deficiency are usually premature for age brain atrophy and delayed myelination of white matter and sometimes small white matter patches.
- In this particular patient, there is no premature for age brain atrophy in both studies and also no delayed in white matter myelination.
- There is no affection of the deep cerebral white matter but affection of the brainstem specially at the posterior aspect of the midbrain and medulla and also affection of the cervical cord and these showed interval regression between the study of 2022 and the study of 2024 as explained above.
- These patches are nonspecific and can be due to primary demyelination like a cases of ADEM or neuromyelitis optica other.
- However, still they can be manifestation of Biotendinase deficiency running regressive pattern, however, due to de-novo occurrence of dorsal cord lesion which needs further cross correlation clinically and laboratory and close follow up as appropriate.

MUCH OBLIGED
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