Indian Journal of Pediatrics
https://doi.org/10.1007/s12098-024-05107-z

CORRESPONDENCE

Amyotrophic Lateral Sclerosis due to *ALS2* Pathogenic Variant Masquerading as Cerebral Palsy: Authors' Reply

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Received: 9 March 2024 / Accepted: 14 March 2024

To the Editor: We appreciate Finsterer J. for critical review and comments on our article published in IJP [1]. Regarding queries raised by the author [2], we would like to highlight our case. The phenotypes associated with *ALS2* variants can be following: Juvenile onset Amyotrophic lateral sclerosis (ALS) type 2 (OMIM#205100), Juvenile primary lateral sclerosis (OMIM#606353) and Infantile onset ascending spastic paraparesis (OMIM#607225) [3]. The clinical features of the above three phenotypes are overlapping as all of them come under the group of Motor Neuron disorders. Even though the gene causes 3 phenotypes, our case had ALS phenotype as there were signs of denervation on electromyography, hence reporting as ALS.

We could not test the parents for the variant due to cost constraints; however, both the parents were asymptomatic clinically. We do not have objective evidence of Apgar score, but child was apparently well with smooth fetal to neonatal transition phase. Child did not require neonatal intensive care unit support immediately after delivery. As per history, encephalopathy was present in the index case in the form of lethargy and poor feeding. There were no objective grading or scaling documents available to diagnose or grade encephalopathy. Encephalopathy did not last more than 5–6 d based on history given by parents. Probable cause was kept as sepsis or hypoglycemia based on historical points given by caretakers. We have undertaken extensive investigations which includes metabolic testing by tandem mass spectrometry, urinary organic acids, serum lactate and ammonia, arterial blood gases to exclude other causes, which were all normal. Hence based on clinical phenotype and investigations, we diagnosed the case as ALS.

Declarations

Conflict of Interest None.

References

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Published online: 23 March 2024