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Leigh syndrome in a Filipino child: A case report

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Introduction: Leigh disease and Leigh-like syndrome are a heterogenous group of neurodegenerative disorder involving any level of the neuraxis and may present with a variety of clinical presentations, prominent among them is psychomotor regression. Despite the remarkable number of established disease genes and novel mutations being discovered, many cases of Leigh syndrome remain without a genetic diagnosis, indicating that there are still more disease genes to be identified.

Case: Here we present a case of a two and a half-year-old girl who presented with delayed acquisition of developmental milestones with subsequent regression, ataxia and dyskinesia. Her work-up show raised blood lactate levels and lactate peak in MR Spectroscopy. Mitochondria genome show absence of mitochondrial DNA mutation, while whole exome sequence analysis reveal a novel dynein gene variant, p.A1577S. Her parents underwent genetic testing as well, and her father also has the same dynein mutation, however is non-symptomatic. She had an older brother who initially presented with ophthalmoplegia and eventually developed psychomotor regression with lactate doublet by MR Spectroscopy. He subsequently expired from respiratory failure after almost 2 years from initial presentation. Both siblings were diagnosed with Leigh syndrome.

Conclusion: The diagnosis of Leigh syndrome remains based on characteristic clinical and radiologic findings. However, a specific defect must be identified if reliable genetic counseling is to be provided.

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