SEPIAPTERINE REDUCTASE DEFICIENCY:
Case Series of An African Descent Family Living in Indonesia

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BACKGROUND

Sepiapterin reductase deficiency (SRD) is an inborn error of metabolism which causes monoamine neurotransmitter deficiency. Symptoms range from developmental delay in infants to movement disorder in older children. It responds well with dopa replacement therapy. We report three cases of SRD in which diagnosis of the first case led to diagnosis of oldest and youngest children in the family.

RESULTS

In this study, the initial symptoms of SRD appeared within the first year of life (ranged from 2 until 6 months) with manifestation of delay motor development and hypotonia. Interestingly, oculogyric crises, sleep disturbance, and autonomic dysfunction, in contrast with previous cases, were absent in the subjects. All patients showed dramatic improvement after treated with L-Dopa/Carbidopa.

CONCLUSION

Early detection and management of SRD lead to improvement of developmental delay and movement disorder, even in older children.