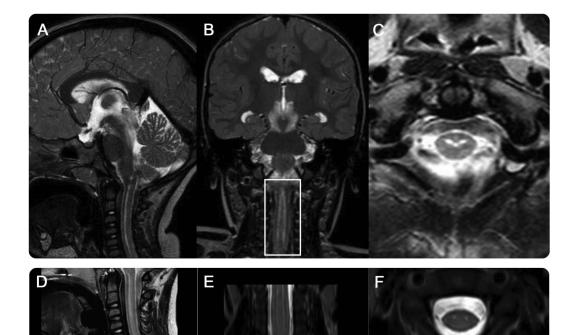


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Teaching Neuro *Images*: Spinal cord gray matter involvement in complex I deficiency mitochondriopathy

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Correspondence to Dr. Bartolini: emanuele.bartolini@meyer.it Figure Brain and cervical MRI at symptoms onset (A-C) and 7 months later (D-F)



(A) Sagittal image shows T2 hyperintensities in posterior midbrain, medulla, and central spinal cord. (B) Coronal image depicts symmetric T2 hyperintensities in subthalamus, midbrain, and spinal cord (white box). (C) Axial image at C1 level (odontoid process of the axis) shows symmetric focal T2 hyperintensities in anterior spinal cord, involving anterior horns and gray commissure. (D) Sagittal image obtained 7 months later shows persistent T2 hyperintensity of central spinal cord. (E) Coronal curved reconstruction and (F) native axial images demonstrate symmetric T2 hyperintensity circumscribed to anterior horns of cord GM.

A 5-year-old boy presented with progressive bilateral ptosis, ophthalmoplegia, spastic paraparesis, and intention tremor/dysmetria. MRI showed symmetric T2-hyperintensities in diencephalon, brainstem, and spinal gray matter (GM) (figure, A–C). Muscle biopsy disclosed increased citrate synthetase (16.32; normal <10.9) and 31% decrease of complex I activity. Mitochondrial DNA and *SURF1* gene sequencing were unrevealing. Leigh-like syndrome was diagnosed based upon the association of clinicoradiologic findings and complex I deficiency. After 7 months on ubidecarenone, thiamine, riboflavin, and carnitine, ptosis and

dysmetria persisted, paraparesis remitted, and T2 hyperintensities decreased (figure, D–F).

In Leigh's original description, spinal cord white matter (WM) was affected. Spinal GM involvement has only been described in association with spinal WM lesions.

Our observation suggests a possible selective involvement of spinal GM.^{1,2}

AUTHOR CONTRIBUTIONS

Case recruitment and clinical follow-up: E. Procopio. MRI acquisitions and image interpretation: M. Mascalchi, E. Bartolini, A. Bianchi, P. Gulino. Drafting of the manuscript: M. Mascalchi, E. Bartolini.

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REFERENCES

- Baertling F, Rodenburg RJ, Schaper J, et al. A guide to diagnosis and treatment of Leigh syndrome. J Neurol Neurosurg Psychiatry 2014;85: 257–265
- Huntsman RJ, Sinclair DB, Bhargava R, et al. Atypical presentations of Leigh syndrome: a case series and review. Pediatr Neurol 2005;32:334–340.



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