■ Clinical commentary

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Epileptic Disorders

ITPA-associated developmental and epileptic encephalopathy: characteristic neuroradiological features with novel clinical and biochemical findings

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Epileptic Disorders

- Mutations in the *ITPA* gene have been recently identified as a cause of developmental and epileptic encephalopathy (DEE)
- The clinical spectrum of *ITPA*-associated DEE is still evolving
- Neuroradiological features are characteristic and aid in suspecting this condition, although the underlying pathophysiology is unclear

We describe two patients with some new clinical findings as well as biochemical changes that could possibly explain the neuroradiological changes in *ITPA*-associated DEE

Epileptic Disorders

Novel findings

- Movement disorders, especially oculogyria and dystonic tremor, as seen in one of our patients, have not been described for this entity before. The oculogyria showed a good response to levodopa.

- Well-controlled seizures, in particular, with good response to levetiracetam, were seen in both patients. All previously reported patients have had drug-resistant seizures.

- One of our patients also had a high CSF glycine level. Glycine encephalopathy was therefore considered a close differential in this patient in view of clinical, biochemical and neuroradiological features, but was refuted based on genetic testing.