

# Lethal neonatal spasticity-epileptic encephalopathy syndrome associated with BRAT1-Mutation

## Aim

Report a rare case of a lethal neonatal spasticity-epileptic encephalopathy syndrome associated with a BRAT1 gene mutation.

## Methods

- Comprehensive clinical assessment and monitoring of the infant's condition.
- Multidisciplinary approach involving neonatology, neurology, genetics, and palliative care.
- Extensive diagnostic work-up including brain MRI, metabolic testing, and genetic mutation screening.

# Results and discussion

- The neonate presented with reduced vigilance, muscle spasticity, weak sucking reflex, coarse facial features, and tonic-clonic seizures at birth.
- Closed eyes without any response on stimulation.
- Despite triple anticonvulsant therapy, there was no reduction in seizure burden.
- A homozygous mutation in the BRAT-1 gene was identified, leading to the diagnosis of BRAT1-associated neurodegenerative disorder.
- This disease is a rare autosomal recessive neurodegenerative disorder that often begins in utero and typically results in death in infancy.

# Conclusion

- Neonatal spasticity-epileptic encephalopathy syndrome is a rare autosomal recessive neurodegenerative disorder caused by BRAT-1 mutation.
- Early recognition and interdisciplinary approach are crucial for timely diagnosis and management.
- BRAT-1 mutations are not routinely screened for in prenatal diagnostics, highlighting the importance of genetic testing in cases of unclear neonatal encephalopathy.
- Poor prognosis necessitates early discussion with parents regarding treatment options and palliative care.

# Authors & affiliation

Vanessa Maibach (1), Cornelia Hagmann (1), Rebecca Fierling (1), Seval Demirbas (2), Andrea Rüegger (2), Eva Bergsträsser (3)

- 1) Department of Pediatric Intensive Care Medicine and Neonatology, University Childrens' Hospital Zurich, Zurich, Switzerland
- 2) Department of Neuropediatrics, University Childrens' Hospital Zurich, Zurich, Switzerland
- 3) Department of Pediatric Palliative Care, University Childrens' Hospital Zurich, Zurich, Switzerland