Pyridoxine-dependent epilepsy (PDE)

- Rare autosomal recessive epileptic encephalopathy
- Antiquitin (ALDH7A1) gene defect affecting lysine catabolism
- Treatment: Life-long pyridoxine supplementation and dietary lysine-reduction

Lysine catabolic pathway in PDE

Methods

- **Objective:** To study clinical and genetic characteristics, neurological outcomes of children with PDE
- **Study type:** Both prospective and retrospective
- **Study population:** Consecutive children with genetically-confirmed PDE
- **Study setting:** Pediatric Neurology Clinic of a tertiary care hospital
- **EEG and MRI**
- **Developmental and cognitive assessment** by Developmental profile-3 (DP-3), VSMs (Vineland social maturity scale)

Results

- **N=8 children**
- Median age=3.37 years (IQR=9.3-1)
- Median age at onset of seizure: day 1 of life (IQR=2.5-1)
- Most common seizure type: Focal-onset
- **EEG:** abnormal in all 8 cases
- Mean age of start of pyridoxine: 31 days
- **Exome analysis:** homozygous (n=6), compound heterozygous mutation (n=2)

Conclusion

- **PDE occurs early in life and pyridoxine can significantly control seizure especially in children with no structural abnormality in brain**
- **Neurological outcomes are subnormal despite early supplementation**
- **Hydrocephalus is an important additional neuroimaging finding**