Child with Late Infantile Batten Disease
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Topic – Child with Late Infantile Batten Disease

Introduction
- Febrile seizures (FS) are common in childhood
- Most often this is secondary to an intercurrent illness
- It is important to take a developmental history in children with FS
- Indication of any developmental regression should trigger further investigations to determine the cause
- Presented here is a case of Late Infantile Batten’s disease whose initial presentation was like FS

Case study
- 3½ year male child presented with seizures and drop attacks; initially diagnosed as febrile seizures
- He had developmental regression
- MRI and EEG were abnormal
- Uncontrolled epilepsy on different anticonvulsants
- Buffy coat electronic microscopy: typical curvilinear inclusions
- White cell enzyme: reduced TPP1 (Tri-peptidase-1)
- Diagnosis: Late Infantile Batten’s disease
- Management: Supportive therapy with multi-disciplinary team involvement; plan – ketogenic diet

Discussion
- Batten’s diseases is a form of Neuronal Ceroid Lipofuscinoses (NCLs)
- 4 types of Batten’s disease has been reported
- Affects 1:30000 births, pan-ethnic, both sexes
- Autosomal recessive condition, late infantile Batten’s disease presents between 2–4 years of age
- Born apparently healthy, develop epilepsy, lose their sight, speech, mental and motor abilities
- Mutation in the CLN2 gene causes a TPP1 enzyme
- Increased deposition of ceroid lipofuscin in brain cells causes seizures and developmental regression
- Management: supportive, seizure control
- Outcome: death occurs between 5 – 14 years

References
1) BDFA (The Batten Disease Family Association – for families, professionals and facilitating research). Available at http://www.bdfa.uk.co.uk/index.html
4) NINDS Batten Disease Information Page. Available at http://www.ninds.nih.gov/disorders/batten/batten.htm