Newcastle Mitochondrial Centre

At a glance guidelines:

Epilepsy in Adult Mitochondrial Disease

There are many different forms of mitochondrial disease, varying greatly in their genetic basis, clinical presentation, progression and prognosis. We recommend referral to a specialist mitochondrial centre for appropriate counselling and guidance (www.mitochondrialncg.nhs.uk or see appendix for international centres).

Epilepsy may be a prominent feature of some forms of mitochondrial disease. We therefore recommend the following:

- 1. The diagnosis of epilepsy is best made by a neurologist or other epilepsy specialist as recommended in the SIGN guidelines.
- 2. Classification: the seizure type(s) and epilepsy syndrome should be identified if possible and may influence the choice of anti epileptic drug (AED).
- 3. Epilepsy in patients with mitochondrial disease should be treated actively from the outset, in order to minimize subsequent cerebral damage related to frequent or intractable seizures.
- 4. Non-convulsive status epilepticus should be actively excluded in the obtunded patient.
- 5. Potential precipitants (eg infection, dehydration, metabolic derangements) should be sought and treated.
- 6. Comorbidities (eg cardiomyopathy, ileus) should be considered, particularly in the ITU patient.
- 7. Sodium valproate should be avoided where possible and especially in patients who might carry POLG mutations due to potential fulminant hepatoxicity.