Synonyms: neuronal ceroid lipofuscinosis juvenile type, JNCL, Spielmeyer-Vogt syndrome

This is one of a group of childhood autosomal recessive lysosomal disorders with profound degenerative effects on the central nervous system.

It was first described by Batten (a British paediatrician) in 1903.[1]

Pathogenesis

It is caused by a mutation in the CLN3 gene at gene locus 16p12.1. This group of diseases represents a new class of lysosomal storage disorders:[2]

- Of the nine clinical variants (CLN1-CLN9), six have been genetically identified.[3]
- As well as the CLN3 gene, these include CLN1, CLN2, CLN5, CLN6 and CLN8.

Epidemiology

It is a rare condition with a variable incidence across different countries.

- In Western Germany it has been estimated as occurring in 0.71 per 100,000 live births.[4]
- It occurs in about 1 to 5 cases per 100,000 generally, but in Finland the figure is around 8 per 100,000. There appear to be slightly different mutations across Europe.

Risk factors

As with other rare autosomal recessives, the main risk is consanguineous marriage. If a child is affected, both parents will be carriers. The risk for further children is that:

- 1 in 4 will have the disease
- 2 in 4 will be normal but carriers of the gene
- 1 in 4 will be normal and not a carrier

Presentation

Onset is between 5 and 10 years old:

- There is often rapid deterioration of vision and a slower, but progressive deterioration of intellect.
- Seizures and psychosis develop later.
- There may be features of Parkinson's disease.
- In some cases, the early signs are subtle, taking the form of personality and behavioural changes, slow learning, clumsiness, or stumbling.

Examination

The fundi show pigmentary degeneration rather similar to retinitis pigmentosa. Referral to an ophthalmologist is useful as abnormal findings may be noted on funduscopy examination, and fluorescein angiography.

Investigation

The pathological features are severe, widespread neuronal degeneration resulting in simple retinal atrophy and in massive loss of brain substance and accumulation of lipofuscin in neuronal perikaryon. There are a number of useful tests:
There is a deficiency of leukocyte peroxidase. This might also be useful in detecting heterozygotes. Both homozygotes and heterozygotes can be identified on the basis of metachromasia in skin fibroblasts in cell culture. Polyacrylamide gel electrophoresis (PAGE) shows low molecular weight peptides in the urine and this may be a specific marker. PAGE can also be used on chorionic villous samples for pre-natal diagnosis. MRI shows general brain atrophy, more in the cerebrum than the cerebellum. Density is reduced in the thalami. If MRI spectroscopy is available it will show almost complete loss of N-acetylaspartate, reduction in creatine- and choline-containing compounds, raised levels of myoinositol and raised lactate in both grey and white areas. Direct gene analysis has been used for antenatal diagnosis.

**Management**

There is little that can be done for the child, but genetic counselling for the parents is essential.

- Anticonvulsants will help the management of epilepsy in children and adolescents.
- If there are features of Parkinsonism, L-DOPA seems to be of benefit, but not selegeline.

Other treatments are of no proven value. Vitamin E, other antioxidants and selenium are all without benefit and, in the few cases where bone marrow transplantation has been tried, there has been no benefit. However, there is the prospect of new therapies following use of allogeneic hematopoietic stem cell transplantation. Normal enzymatic activity has been reported, without the need for any medication. Reconstruction of the central nervous system has also been reported. However numbers of treated patients are small and further research is needed on safety.

**Prognosis**

There will be mental impairment, worsening seizures and progressive loss of sight and motor skills. Batten's syndrome is often fatal by the late teens or twenties.

**Further reading & references**

- Batten Disease Family Association Website
- Chang CH; Neuronal Ceroid Lipofuscinoses. eMedicine, Sept 2009.

5. Ceroid Lipofuscinosis; CLN3, Online Mendelian Inheritance in Man (OMIM)

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