SYNDROMES OF CHILDHOOD & ADOLESCENCE IN WHICH EPILEPSY MAY OCCUR

AICARDI’S SYNDROME

Aicardi’s Syndrome (AS) is a rare syndrome which occurs only in girls due to an abnormality in the way the brain has developed before birth, probably before the 8th week of pregnancy. At present ante-natal diagnosis is not possible.

As well as this general abnormality of the brain there are three specific features of the syndrome which are:

1. Absence of the corpus callosum (large group of nerves passing from one side of the brain to the other
2. Abnormalities of the retina (back of the eyes)
3. The development of infantile spasms

Sometimes other abnormalities can occur – in the bones of the spine, in the heart and in the facial features.

SYMPTOMS

The first problem that a child with AS generally develops is epilepsy. This always starts in the first six months of life, sometimes shortly after birth.

The first seizures are usually infantile spasms (also called salaam attacks), which involve the arms going stiff and the legs bending so the child’s knees move towards the stomach. Sometimes the arms are flung outwards suddenly and the back may arch. Only one side of the body may be involved. Other types of epileptic seizures may also occur including partial, tonic-clonic seizures or absences. Many seizures can occur in the course of a day.

Delayed development usually becomes apparent from an early age. Children are generally very floppy but sometimes can be very stiff.

Learning difficulties are always present and are usually severe with little social development.

From an early age abnormal eye movements and other eye abnormalities may be noticed. However, the specific abnormality of the eye in AS is not apparent unless the back of the eye (retina) is examined with a special instrument – an ophthalmoscope. This examination will show multiple abnormalities of the retina called lacunae which are due to the abnormal development of the retina. These abnormalities are specific to AS and are not thought to occur in any other condition. As a result of these abnormalities vision is usually impaired but total blindness is rare.

DIAGNOSIS

Diagnosis of AS is usually made by examination of the eyes, a CT or MRI Scan, which provides an X-ray of the head, and an EEG (electroencephalogram) test which records the ongoing electrical activity in the brain.
TREATMENT

There is no specific treatment for AS. Anti-epileptic drugs will be used to try to control the seizures. A number of different antiepileptic drugs may have to be tried to control the seizures, but in many cases achieving control may prove to be very difficult.

Managing the child with AS involves controlling their seizures with drugs and providing the support for the child and the family that is always needed in such cases of severe, multiple disabilities. Such support involves access to the most appropriate educational placement, physiotherapy, occupational therapy, and the provision of aids for the child as well as the appropriate emotional and practical support for both the child and the family.

PROGNOSIS

The long term outlook for children with AS is poor with persisting seizures and severe learning difficulties. Children with AS have a reduced life expectancy due to the severe seizures, the extent of their physical disability and their susceptibility to chest infections. Some children with AS may survive into adolescence or early adulthood.

This condition does not run in families and there is virtually no risk of it happening again in the same family.