



Aicardi syndrome

Epilepsy Action is indebted to Dr Richard Appleton, a consultant who specialises in children's epilepsy, and his associates, at Alder Hey Hospital, Liverpool, who have kindly written this fact sheet.

For further information about epilepsy or anything mentioned in this factsheet, please contact the Epilepsy Helpline freephone 0808 800 5050 or helpline@epilepsy.org.uk.

What is a syndrome?

A syndrome is a group of signs and symptoms that, added together, suggest a particular medical condition. In epilepsy, examples of these signs and symptoms would be things like the age at which seizures begin, the type of seizures, whether the child is male or female and whether they experience difficulties with learning.

Aicardi syndrome

This is a very rare syndrome which only occurs in girls. It is thought the syndrome causes a male unborn baby to be lost as a miscarriage. Affected girls will have developmental delay, seizures, structural brain abnormalities including a partially or completely absent corpus callosum (the structure that links the two halves of the brain together). They will also have very typical eye abnormalities which often help to make the diagnosis. The abnormalities on the retina at the back of the eye are called choroidal lacunae. They appear as distinctive, round 'footprint shaped' yellow-white lesions (mark) visible with a special

piece of equipment called an ophthalmoscope. As well as an absent corpus callosum, some babies may have brain cysts or other brain abnormalities present. These will be detected with a magnetic resonance image (MRI) scan. Other features of Aicardi Syndrome include cleft lip and palate, asymmetry of the face (lopsided, uneven), microcephaly (a small head), abnormally formed bones in the backbone and scoliosis (curvature of the back). Aicardi syndrome does not appear to run in families. It is thought to be caused by a spontaneous mutation (random change) in the X chromosome (gene) which happens at conception.

Symptoms

Seizures often happen before three months of age and take the form of infantile spasms (West syndrome*). Less commonly, babies will have other seizure types including partial motor seizures and complex partial seizures. Seizures are often resistant to anti-epileptic medication. The electroencephalogram (EEG) will often show the pattern found in West syndrome which is known as hypsarrhythmia. Rarely the EEG may show abnormal discharges or spikes and waves alternating with periods of flatness coming independently from each side of the brain (independent burst suppression pattern).

Diagnosis

The diagnosis may not be made until the affected girl develops seizures and investigations such as an MRI scan and a special eye exam are carried out.

Treatment

The choice of medication is difficult as seizures are often resistant to medication. Depending on the type of seizures present, drugs which may be effective include vigabatrin (Sabril) or a course of corticosteroids.

Prognosis (outlook)

All girls with Aicardi syndrome will have moderate or severe learning difficulties. They will remain dependent on others for all of their life. They may have problems with feeding, and may develop stiffness (spasticity) in their arms and legs. Rarely some girls may learn to walk and develop some limited speech. They frequently have complications such as chest infections which may shorten their life.

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* Factsheet on West syndrome is available from Epilepsy Action.

For information on seizure types, anti-epileptic medication and general aspects of epilepsy please contact the Epilepsy Helpline freephone 0808 800 5050.

Other possible contact for support is Contact a Family, 209-211 City Road, London, EC1V 1JN, telephone 0808 808 3555, www.cafamily.org.uk

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