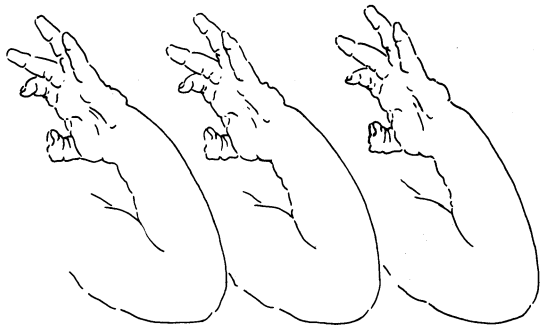
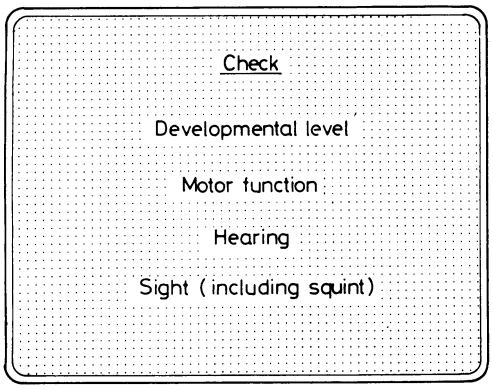
ABC of 1 to 7

H B VALMAN

EPILEPSY



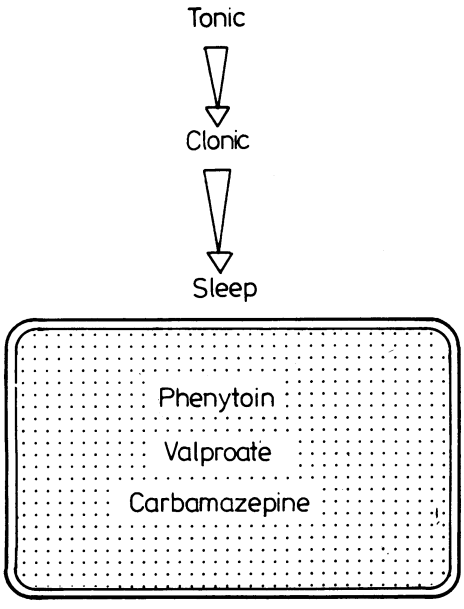


Repetitive attacks with similar features are essential for the diagnosis of epilepsy. The attacks may cause changes of consciousness or mood or produce abnormal sensory, motor, or visceral symptoms or signs. These changes are caused by recurring excessive neuronal discharges in the brain, although the electroencephalogram (EEG) may be normal. Investigations are no substitute for a history taken carefully from a witness. Documented absence of fever is essential to exclude the more common problem of febrile convulsions (see *next* article). The incidence of epilepsy is about 6 in 1000 schoolchildren compared with the incidence of children with febrile convulsions, which is about 30 per 1000 preschool children.

Disability depends partly on the frequency and severity of the fits but also on the presence or absence of developmental delay, cerebral palsy or defects in the special senses which would suggest brain damage. The word "damage" will be used in this paper, but for discussion with parents "abnormality" is preferred. Most children with epilepsy attend normal schools, rarely have fits and have no disability apart from the fits.

Epileptic fits can be divided into generalised or partial seizures. Generalised seizures include grand and petit mal and myoclonic fits. Partial seizures include focal and temporal lobe fits.

Grand mal

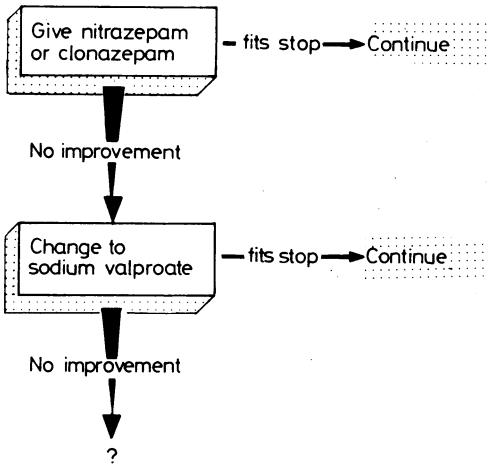


About 80% of children with epilepsy have grand mal (tonic-clonic) seizures. The child may appear irritable or show other unusual behaviour for a few minutes or even for hours before an attack. Sudden loss of consciousness occurs during the tonic phase, which lasts 20-30 seconds and is accompanied by temporary cessation of respiratory movements and central cyanosis. The clonic phase follows and there are jerking movements of limbs and face. The movements gradually stop and the child may sleep for a few minutes before waking, confused and irritable. The best prognosis occurs in older children and those who respond promptly to anticonvulsants. When epilepsy is secondary to brain damage the prognosis may be less good. Phenytoin and valproate are the commonly used drugs. Carbamazepine has special value in children with brain damage. Anticonvulsants are given until two to four years have passed with no symptoms and then discontinued gradually over several months.

Petit mal

In petit mal episodes of altered consciousness lasting 10-15 seconds occur spontaneously and can be precipitated by hyperventilation. Petit mal is rarely associated with mental retardation or brain damage. There is a typical EEG appearance, and the frequent attacks respond promptly to ethosuximide or to sodium valproate introduced slowly. Treatment is continued for two years after the fits have been controlled.

Myoclonic epilepsy

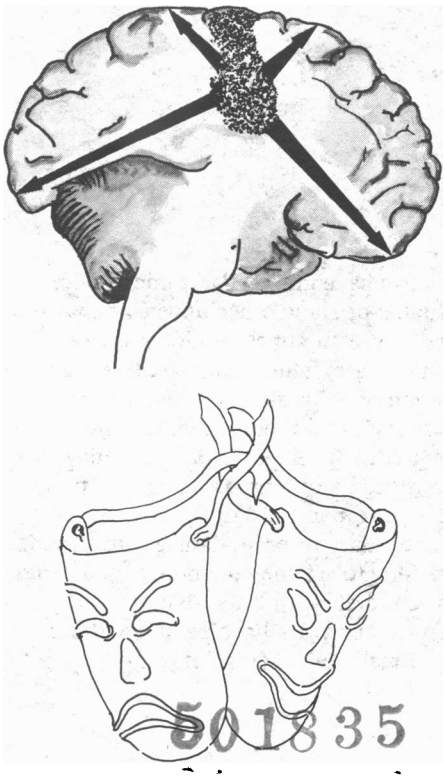


Myoclonic epilepsy is caused by different brain insults; heredity may be implicated. Many of these children have developmental delay and some evidence of brain abnormality before the fits begin. The child may have a variety of seizures including (*a*) symmetrical synchronous flexion movements (myoclonic); (*b*) brief loss of consciousness; and (*c*) sudden head-dropping attacks (atonic-akinetic). Infantile spasms are a form of myoclonic epilepsy which starts before the age of 1 year and have a characteristic EEG.

Perinatal asphyxia or acquired brain damage from any cause may have been present. Many of the children have mental retardation but the degree is variable. The EEG may remain normal long after the onset of the symptoms. Myoclonic epilepsy must be distinguished from petit mal epilepsy as treatment and prognosis are different.

Myoclonic seizures are often difficult to control with drugs. Nitrazepam or clonazepam is introduced gradually until the attacks cease or drowsiness occurs. Sodium valproate is usually the next drug used. As a last resort it may be necessary to use a combination of various drugs or a ketogenic diet.

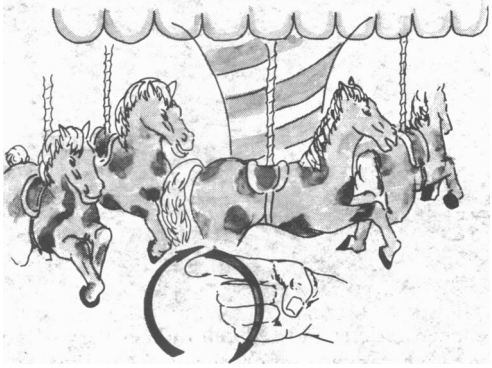
Partial seizures



Partial seizures originate in specific areas of the brain and the symptoms depend on the site of the epileptic focus. Perinatal asphyxia or prolonged "complex" febrile convulsions are the main cause; a progressive space-occupying lesion is an extremely rare cause of this clinical picture. The commonest variety of partial epilepsy in childhood is benign focal epilepsy of childhood where the focus is in the Rolandic area. It usually starts between the ages of 7 and 10 years and attacks begin especially during sleep. Often they become generalised so that any generalised nocturnal convulsions may be due to this condition, which has a good prognosis. Consciousness is often retained but the child does not speak or swallow during the attack. There may be jerking of one side of the face with salivation, gurgling noises, and peculiar sensations affecting the tongue. Carbamazepine is extremely effective and most of the children are completely free of fits and then need no drugs shortly after puberty.

In contrast, the great variety of bizarre symptoms produced by fits originating in the temporal lobe makes diagnosis difficult and attacks may be intractable despite anticonvulsants. There may be short episodes of emotional disturbance with the sudden onset of fear or rage, hallucinations of sight, sound, or smell or visceral symptoms such as epigastric discomfort. A generalised tonic-clonic seizure may follow in some children. Carbamazepine is effective in about half the patients and is introduced slowly over several weeks to avoid drowsiness and ataxia. Phenytoin is the next drug to be tried. Rarely surgical removal of the temporal lobe has been performed and is effective in about 50% of children when medical treatment has failed to control the fits. The best surgical results occur when histology shows mesial temporal sclerosis, which follows prolonged "complex" febrile convulsions.

Differential diagnosis

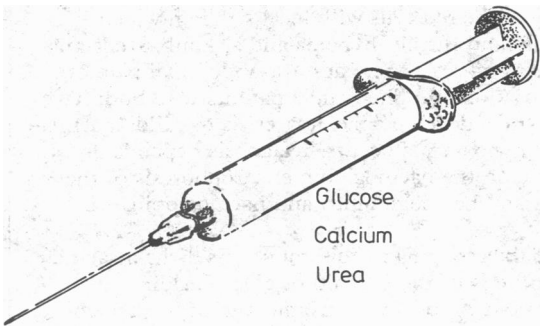


Breath holding attacks—Convulsions need to be differentiated from breath-holding attacks, which usually begin at 9 to 18 months. Immediately after a frustrating or painful experience the child cries vigorously and then suddenly holds his breath, becomes cyanosed, and in the most severe cases loses consciousness. Rarely his limbs become rigid, and there may be a few clonic movements lasting a few seconds. He takes a deep breath and regains consciousness immediately. The attacks diminish with age and there is no specific treatment. Mothers may be helped to manage these extremely frightening episodes by being told that the child will not die and that they should handle each attack consistently by putting the child down on his side.

Syncope or a faint may occur at any age but is more usual in older children. While in the upright position the child appears very pale, becomes unsteady, and falls to the ground. There may be a precipitating factor such as standing in one position for a long time or being in a closed, hot room. Rarely there may be a few clonic movements of the limbs but never a generalised convulsion and within a few minutes the child is perfectly normal again. He may say that he felt dizzy or unsteady at the beginning of the attack. Isolated episodes with obvious precipitating factors require no treatment.

Acute labyrinthitis is another cause of episodes of dizziness. If the child is asked to draw his sensation in the air with his finger he will describe a circular movement which suggests vertigo. This is due to a viral infection affecting the balance mechanism of the inner ear which usually resolves within a few weeks, although attacks occasionally persist for longer.

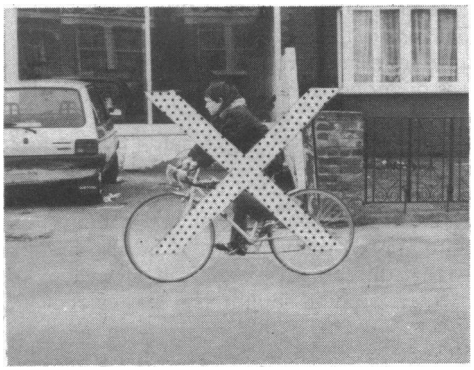
Investigations



Investigations should be performed as outpatient procedures, keeping them to the minimum necessary for making a firm diagnosis and for excluding treatable causes. The specific tests will depend on the diagnosis made after taking the history and examining the child, but most children will have blood taken for estimating fasting plasma glucose, calcium, and urea concentrations. A Dextrostix test should be performed during a fit and if abnormal, blood is taken for a blood glucose estimation.

Radiology of the skull may show abnormal calcification. The EEG should not be used to determine whether a child has epilepsy; this is a clinical decision. But the EEG may provide guidance on the type of epilepsy so that appropriate drugs are given, or it may show a unilateral lesion indicating the need for a brain scan by computerised axial tomography. The EEG does not show whether the epilepsy is improving or whether it is safe to stop treatment.

General management



Regular measurements of blood or salivary anticonvulsant levels may help to prevent side effects and confirm compliance but the dose must be determined mainly by the presence or absence of fits. Children with epilepsy should not ride a bicycle in the open road or swim unless there is an adult with them in the water. They should not climb ropes or high bars in a gymnasium. They can carry out all other activities. The school teacher needs to know the child's diagnosis and be aware that most children with epilepsy have normal intelligence and should be expected to perform as well as their peers.

2 + 3 = 6

5 - 1 = 3

1. Anticonvulsant blood level ↑
2. Fits ++
3. Intelligence ↓

Learning difficulties may be due to the effects of anticonvulsants, inattention caused by unrecognised fits or underlying brain damage. Epilepsy is a family problem which can modify the lives of all members and the parents will be worried about the child's prospects for future employment, driving a car, and marriage. They may believe, wrongly, that epilepsy is always associated with mental retardation. The doctor should tell the parents that the fits are *not* due to a tumour, that the child will *not* die in an attack, and that a short fit does *not* injure the brain.

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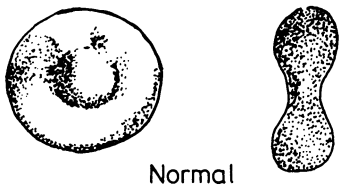
Normal haematological values and diagnosis of haemoglobinopathies

Normal ranges		
	Haemoglobin (g/dl)	Mean corpuscular volume (fl)
<1 years	10·5 - 13	70-84
1-4 years	11·5 - 13·5	74 - 86
4 - 7 years	11·5 - 13·5	76-86
Adult	13 - 15	80-100
Mean corpuscular haemoglobin concentration is constant throughout life (30-34 g/dl)		



Normal

Thalassaemia

Sickle cell

Normal ranges for haemoglobin concentrations and red cell indices vary with age. The normal ranges printed on standard report forms are derived from adult studies and their use for children may result in a wrong diagnosis, especially of iron deficiency anaemia.

The child with severe anaemia due to sickle cell anaemia thalassaemia major receives an abnormal gene from each parent, who has no symptoms. In sickle cell anaemia there is a chronic haemolytic anaemia with superimposed crises due to local sickling, marrow aplasia, or acute haemolysis. Hypoxaemia causes deformity of the red cells with local sickling, which results in blocking of capillaries and further hypoxia and sickling. Ischaemia distal to the local sickling lesion causes bone, chest, or abdominal pain or infarcts in the brain, spleen, or kidneys. The child's parents who both have the trait (or a child with the trait) develop a sickling crisis or mild anaemia only if very severe prolonged hypoxaemia occurs. Diagnosis depends on detecting haemoglobin S alone on the haemoglobin electrophoresis of the child with sickle cell anaemia and haemoglobin S and haemoglobin A in both parents.

The most common type of thalassaemia in this country is β-thalassaemia, in which there is reduced synthesis of the β-chain of globin leading to reduced synthesis of haemoglobin A and hypochromic microcytic anaemia. Synthesis of haemoglobin F and haemoglobin A_2 are not affected and so the percentages of haemoglobin F and A_2 are increased.

Two genes for thalassaemia are present in a child with thalassaemia major and one in those with thalassaemia minor. The child with thalassaemia major has severe chronic haemolytic anaemia with a very large liver and spleen and later enlargement of the frontal and malar bones due to bone marrow hyperplasia. He usually needs regular blood transfusions throughout his life. Thalassaemia minor causes mild or no anaemia. The blood picture is similar to that of iron deficiency anaemia but the mean cell volume (MCV) is disproportionately reduced compared with the red cell count (RBC) and haemoglobin levels; the ratio MCV/RBC is less than 12 and the serum iron level is normal. The diagnosis is confirmed by finding a raised percentage of haemoglobin A_2 or haemoglobin F, or both.

If thalassaemia minor is suspected during a routine antenatal clinic blood count, the father should also be tested immediately. If he also has thalassaemia minor, the possibility that the fetus has thalassaemia major should be considered and further expert opinion sought as a matter of urgency.