True Myoclonic Epilepsy in Childhood

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The term 'myoclonic epilepsy' is used in adults to describe a form of epilepsy in which a sudden, involuntary, and momentary contraction occurs in a single muscle or muscle group, often without apparent loss of consciousness. Such attacks may occur in isolation, or as part of the pattern of epilepsy in patients experiencing *grand mal* or other forms of epilepsy. A recent authoritative review of the subject in adults is given by Aigner and Mulder (1960).

It is generally agreed that myoclonic epilepsy does occur in childhood, but confusion has arisen on account of differing descriptions and nomenclature. Early writers such as Unverricht (1891) and Lundborg (1903) used the term myoclonus to denote any involuntary movement or muscle jerk occurring during a degenerative neurological illness, and considered myoclonus to be a symptom of underlying organic disease. Later the term 'myoclonic' was used by many authors to describe the involuntary muscle jerks that occur as subsidiary phenomena in the intervals between *grand mal* attacks in patients with major epilepsy. Muskens (1909) described 'the common occurrence of single, sudden and often isolated convulsions (or regional jerks) with sudden adduction or abduction of one or both arms, or a sudden flexion of extension of the trunk'. He considered that these episodes occurred most often in intervals between *grand mal* attacks, or that they might precede the onset of *grand mal* by as much as several years.

Hodskins and Yakovlev (1930) found myoclonus in 10-15% of a mixed age-group of 300 cases of all forms of epilepsy, and commented upon a frequency of a positive family history of epilepsy and a high incidence of cerebellar signs in patients showing myoclonus. Gastaut (1954) commented that myoclonus had become a somewhat neglected symptom in most books on epilepsy and deserved to be reinstated. Bridge (1949) recognized the occurrence of myoclonic epilepsy in children, though he considered this to be a very rare form of epilepsy in childhood.

In recent years, discussion of myoclonic epilepsy in childhood has become confused by the increasing tendency for the term 'myoclonic' to be attached to the syndrome of infantile spasms with hypsarrhythmia and subnormality, such terms as 'myoclonic infantile epilepsy' (Brandt and Melchior, 1960) and 'myoclonic encephalopathy of infants' (Kinsbourne, 1962) being typical of many similar titles used. As will be shown in this paper, true myoclonic epilepsy in childhood differs in many respects from the infantile spasms syndrome, and a clear distinction should, therefore, be made between the two conditions.

It seems reasonable to retain the term *infantile spasms* to describe the syndrome of spasms, hypsarrhythmia, and mental subnormality, and to reserve the term *myoclonic epilepsy* for the type of attack to be described.

Material and Methods

When it was realized that a number of children suffering from myoclonic epilepsy were attending the clinic, the records of the Children's Hospital between 1955 and 1964 were scrutinized, and 14 children were found and recalled for review. Children with infantile spasms and akinetic attacks, misdiagnosed as myoclonic epilepsy, were excluded.

Review included clinical examination and repeat EEG. It was not possible to obtain formal intelligence testing of the children but they were allocated to defined categories according to intellect (see key to Table III).

Clinical details of patients are summarized in Tables I and II. The relevant EEG data appear in Table III.

Clinical Features

The attack. The most striking features of this form of attack are the violence of the muscular contraction and the absence of any warning or aura, so that the child is taken unawares and has no time to prepare himself. The attack is brief, lasting only a fraction of a second, and the post-ictal disturbance is slight. Typically a child in the age-group 3 to 7 years develops this form of attack in which there is a sudden violent contraction of

TABLE I
Clinical Details and Physical Examination on Review

Case No.	Sex	Age (yr.)	Birthweight (kg.)	Perinatal History	Family History of Epilepsy	Recent Clinical Examination
1	M	7½	3·6	Normal	None	Normal
2	F	11½	—	Normal	None	Normal
3	F	8	4·1	Normal	None	Normal
4	F	14	3·1	Normal	None	Normal
5	F	11½	3·1	Normal	None	Normal
6	F	5	2·5	Normal until diarrhoea and dehydration age 7 wk.	None	Mild left hemiparesis
7	M	12	3·75	Normal	None	Normal
8	M	9	2·9	Poor, in incubator several days	None	Normal
9	M	10½	3·4	Normal	Mother <i>petit mal</i> as a child, no other family history	Normal
10	F	18	4·1	Unwell age 3 dy. and developed divergent squint	None	Mild right hemiparesis and rotary nystagmus both eyes
11	M	3½	4·0	Normal	None	Normal
12	M	5½	4·1	Normal	None	Normal
13	F	6½	2·9	Slow to thrive; hypothyroid state diagnosed May 1961	None	Normal
14	M	5	—	Normal	Strong family history of major epilepsy	Normal

Birth history was normal in every case.

TABLE II
Details of Epilepsy Follow-up

Case No.	Age (yr.)	Sex	Age at Onset of Epilepsy (yr.)	Age at Onset of Myoclonic Attacks (yr.)	Whether Consciousness Lost in Myoclonic Attacks	Injuries*	Present Mental Category†	Length of Follow-up (yr.)	Age of Cessation of Attacks (yr.)	Present State of Epilepsy
1	7½	M	2½	3½	No	++	B	5	—	Infrequent fits
2	11½	F	2	2	Momentary only	++	A	9	4	Completely free
3	8	F	2½	2½	None or momentary	+++	B	6	—	Fair
4	14	F	4½	6	None or momentary	+++	C	8	—	Fair
5	11½	F	5½	5½	Not lost	0	A	6	11	Complete control on drugs
6	5	F	4/12	2	Momentary loss only	+++	A	1½	—	Frequent
7	12	M	3½	4	Momentary or not at all	0	A	8	4	Completely free
8	9	M	4	4	None	0	A	5	7	Free
9	10½	M	3	5	Brief but definite loss	0	B	5	—	Infrequent, i.e. one a month
10	18	F	6	6	Momentary or no loss	++	B	10	—	Infrequent
11	3½	M	3/12	8/12	Momentary only	++	A	3	—	Still frequent
12	5½	M	2½	3½	Momentary only	+	A	2	4½	None
13	6½	F	1½	1½	Probably no loss of consciousness	+++	C, but hypothyroid	5	—	Still having infrequent <i>grand mal</i> and myoclonic attacks
14	5	M	3½	4	Momentary unawareness only	+	A	2	5	Free of attacks

* + + +, serious injuries including fractures and severe head and facial injuries requiring protective headgear; ++, Significant injuries; +, minor injury in attacks; 0, no injuries.

† A, intellect entirely unimpaired and attending a normal school; B, slight but definite impairment of intellect, requiring attendance at a special school; C, more severe impairment of intellect, requiring attendance at training or occupational centre. (Note, no cases fell into a lower category of ineducability.)

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TABLE III
EEG Correlation

Case No. and Sex	Present Age (yr.)	No. of Records	Age Span (yr.) of EEGs	Summary of EEG Recordings
1 M	7½	16	2½-7½	The records up to 1963 showed slow-wave abnormality, few spikes, and much artefact; those in Oct. and Nov., 1963 associated with subclinical myoclonic status showed spike and wave mainly around 1-2 c/sec.; from 1964, discharges have been polyrhythmic spike and wave at 2-4 c/sec., sometimes with polyspikes
2 F	11½	27	2-11	Early records show atypical spike and wave activity, with irregular slow wave components varying between 2-6 c/sec.; no abnormality on photic stimulation; EEG normal since age 4 yr.
3 F	8	3	2-8	Irregular spike and wave activity, varying between 2-4 c/sec., with associated polyspikes; no abnormality on photic stimulation; EEG remains abnormal
4 F	14	11	5-14	Atypical spike and wave activity, showing wide variety of frequencies in slow wave component, varying between 1.5-6 c/sec.; no abnormality on photic stimulation; EEG remains abnormal with atypical spike and wave and multiple spike foci
5 F	11½	9	5½-10	Atypical spike and wave varying at intervals over years between 1-5 c/sec.; no abnormality on photic stimulation; EEG remains abnormal in spite of good drug control
6 F	5	5	3-5	Irregular spike and wave discharges from 2.5-6 c/sec.; no polyspikes; photic stimulation not performed; EEG remains abnormal
7 M	12	10	3½-12	Irregular spike and wave activity with polyspikes; on one occasion myoclonic jerk occurred during photic stimulation and was associated with polyspike on EEG; EEG normal in 1958 when fits ceased
8 M	9	5	5-8	Irregular and atypical spike and wave activity between 2-3 c/sec.; no abnormality on photic stimulation; EEG became normal by 7 yr.
9 M	10½	10	5-8½	Irregular and atypical spike and wave activity, mainly at 2 c/sec.; no accentuation on photic stimulation; EEG remains abnormal, with long periods of spike and wave discharges
10 F	18	6	8½-18	EEGs show left-sided focal lesion, which fits in with left porencephalic cyst; over years considerable irregularity in spike and wave discharges, varying between 2-6 c/sec.; EEG remains abnormal
11 M	3½	12	10/12-3	Recordings show irregular spike and wave activity with polyspikes; abnormality on photic stimulation at 15 months; EEG normal between 18 mth. to 2 yr.
12 M	5½	3	3½-5	Initial record normal, but sleep record showed atypical spike and wave activity at 2-5 c/sec., with polyspikes; no abnormality on photic stimulation; most recent record shows 2-5 c/sec. generalized spike and wave activity
13 F	6½	4	6/12-6	Atypical bilateral spike and wave discharges with polyspikes, worse on photic stimulation; recent EEG normal
14 M	5	2	4-5	Atypical slow spike and wave induced on overbreathing on initial record, mainly at 2 c/sec.; recent record within normal limits

the muscles of the neck and trunk, often with an associated jerk of the arms and often followed by a violent fall forward or backwards, with injury.

Parents frequently describe the attacks in dramatic phrases. For example, the boy in Case 1 was described as 'being struck by lightning' in an attack and on occasions would actually jerk off the ground as a preliminary to a severe fall in which he would often injure his head. Another description is that a child who is holding a toy or a spoon in his hand will characteristically throw it for some distance as the muscle contraction occurring in the attack begins. The face may be forcibly jerked down into the plate or cup, and that this possible source of injury worried some parents is shown by the measures adopted by the parents of the girl (Case 2) who bought soft plastic table-ware to protect her face from injury at meals. Several children were described as being 'thrown down as though someone had pushed them', at the onset of an attack.

Precipitating factors. These varied with different children, but attacks occurred most commonly in the morning, or when the child was

tired later in the day. Attacks occurred at night in three children and one of these (Case 12) was shown to be sleep sensitive on EEG. In one child (Case 1), attacks could be precipitated by combing her hair, drinking, and correcting her verbally.

Loss of consciousness. Characteristically, in the attacks described, this was absent or minimal. It is difficult to be sure whether a child is in touch with his environment during an abrupt and severe myoclonic attack, but certainly loss of consciousness is very brief if it occurs at all, and most children appeared quite normal after an attack.

Injuries. Not surprisingly, in view of the violence of the attacks, injuries were common and significant injury occurred in 8 out of 14 children (see Table II). Recurrent injury to the head was a particularly difficult problem but was lessened by protective head-gear (hard riding hats) which was advised for 5 of the children.

Family history. A family history was unusual in the children described, in contrast to earlier

descriptions of myoclonus, e.g. Hodskins and Yakovlev (1930). Only one child in the present series had a strong family history of epilepsy.

Age of onset. The average of onset of the myoclonic attacks was $3\frac{1}{2}$ years. In 9 children the myoclonic attacks were preceded by other forms of epilepsy (usually *grand mal* attacks) for a period ranging between 3 months and 2 years.

Clinical examination. Physical examination was negative in all but two children (Cases 6 and 10), in whom residual neurological signs were the result of known cerebral insults in infancy. Abnormal cerebellar signs were not present in any child.

Prognosis. This is a severe form of epilepsy which is resistant to therapy and disabling when a child is having frequent fits. The prognosis for life and general health is good and there was a tendency towards improvement in the myoclonic attacks with advancing age. Five children were free of attacks at the time of review. There was no intellectual deficit in 8 of the children, and all of the remaining 6 were benefiting from special school facilities.

Drug control. From the study of this series of 14 children, it proves impossible to obtain any accurate estimate of the value of individual drugs in the management of this form of epilepsy. This is mainly due to the fact that their epilepsy was so highly resistant to the standard anticonvulsant drugs that most patients received a wide variety and combination of anticonvulsants before control was achieved. One child (Case 2) responded dramatically and permanently to a ketogenic diet, another (Case 10) became fully controlled for the first time when carbamazepine ('tegretol') was added to her existing anticonvulsants.

No claim could be made for superiority of any other drug used, though diazepam ('valium') was generally of the greatest use in obtaining control of seizures.

All but 3 children were still receiving anti-convulsant therapy at the time of their follow-up examination.

Incidence. True myoclonic epilepsy in children is certainly not common, though it may well be more common than is generally recognized owing to the confusion with cases in the infantile spasms group, as previously mentioned.

Confusion with so-called 'akinetic attacks' may

also account for a failure to recognize some cases of true myoclonic epilepsy, as pointed out by Gastaut (1954).

The incidence previously described by various authors varies widely, and this is not surprising as Muskens (1909) and Hodskins and Yakovlev (1930), who reported myoclonic episodes in between 10 to 15% of all cases of epilepsy, were describing all those patients in whom myoclonic attacks occurred as a subsidiary manifestation associated with *grand mal* attacks. Aigner and Mulder (1960) also consider the incidence of myoclonus in association with major epilepsy to be approximately 10% of all cases.

Gastaut (1954) gives the incidence of myoclonic epilepsy as being 0.3% of all forms of epilepsy taken over all age-groups. Bridge (1949) found that in 1 to 2% of his series of children with epilepsy this took the form of myoclonic attacks.

In this present series, 14 children out of 376 new cases of epilepsy admitted to Birmingham Children's Hospital in the 10 years under review suffered from this form of epilepsy, giving a proportion of 3.8% of all cases of epilepsy in this paediatric age-group.

EEG findings in adults. The pattern of the EEG in adult myoclonic epilepsy is well recognized. Gastaut (1954) described myoclonus as being represented by a volley of spikes, often multiple, which he termed a 'polyspike'. This may also occur in association with slow-wave, in which case the complex is termed 'polyspike and wave'. Gastaut found the distribution of these discharges to be bilateral, synchronous, and symmetrical, as is characteristic of generalized epilepsy. Gibbs and Stamps (1958) comment on the association of myoclonic attacks with multiple high voltage spikes, often mixed with slow waves. Of the 142 mainly adult patients with myoclonus described by Aigner and Mulder (1960), EEG data were available in only 42 cases. Of these, 27 showed diffuse atypical spike and wave activity varying between 1 to 4 c/sec. Of these 27 cases (age unstated but predominantly adults), 13 showed EEG sensitivity to photic stimulation.

EEG findings in childhood. Few data have previously been available regarding the EEG pattern of true myoclonic epilepsy in childhood. The EEG abnormalities shown in the 14 patients presented here are summarized in Table III, but are briefly described as follows

All children showed abnormal EEGs at some time. The majority of records showed atypical spike and wave discharges which were bilateral

FIG. 1.—Case 4, aged 8 years. Polymorphic, polyrhythmic, spike and wave discharge lasting 8 seconds with jerking of eyelids. Compare with Fig. 2 taken a week later.

and of varying duration (see Fig. 1, 2, 3, and 4). Polyspike discharges were common (Fig. 3). The slow components of the discharges varied from 1 to 6 c/sec. and the discharges did not appear to vary

with the patient's age. One might expect to find slower (1 to 2 c/sec.) components below the age of 3 years and 3 to 4 c/sec. components in the older children, but this was not so. A 2 c/sec. component

FIG. 2.—Case 4, aged 8 years. Spike and wave discharge with 2 c/sec. component dominating. No clinical change observed. The record was taken a week after Fig. 1.

FIG. 3.—Case 11, aged 17 months. Atypical spike and wave discharge, with initial polyspikes and associated myoclonic jerk of head.

was common at all ages and no case showed 'typical' 3 c/sec. spike and wave discharges. No patient showed hypsarrhythmia. There was a great deal of variability in the discharges both from patient to patient and in serial records of the same patient (see Fig. 1 to 4).

In 7 of the patients, myoclonic jerks occurred during the EEG recording. The EEG discharges associated with these jerks were essentially the same as the interictal discharges of atypical spike and wave, though some cases showed some higher amplitude spikes and polyspikes at the time of the

FIG. 4.—Case 5, aged 7 years. Discharge mainly of slow waves at 1.5 to 2 c/sec. with few spikes. Myoclonic jerking of legs and face.

ictus (Fig. 1 and 2). In two of the patients the myoclonic jerks were elicited by photic stimulation. Only 3 cases showed accentuation of spike and wave discharges during photic stimulation, which is in contrast to the figure of nearly 50% of adult patients showing photosensitivity in the series by Aigner and Mulder.

The EEG pattern of the children described here undoubtedly shows a resemblance to that described in cases of 'childhood encephalopathy with diffuse slow spike-waves' or Lennox syndrome (Gastaut, Roger, Soulayrol, Tassinari, Régis, Dravet, Bernard, Pinsard, and Saint-Jean, 1966). The clinical symptomatology in Lennox syndrome consists of 'absences' which are atypical in that the onset and ending of the attack is gradual rather than abrupt. Consciousness is often clouded rather than completely lost. Further, in contrast to true *petit mal*, there is frequently considerable mental retardation. Tonic and clonic seizures also occur in Lennox syndrome but the clinical picture is quite different from that in myoclonic epilepsy, and confusion between the two forms of epilepsy should not occur on clinical grounds.

By contrast, a superficial clinical resemblance to myoclonic attacks may be observed in the 'generalized tonic seizures' described by the same workers (Gastaut, Roger, Ouahchi, Timsit, and Broughton 1963). However, as Gastaut himself points out, the myoclonic attack is very much shorter than the tonic seizure, which usually lasts between 10 and 20 seconds and may last up to 60 seconds. The EEG pattern in these tonic seizures is also entirely different, in that in tonic seizures there is commonly an initial period of flattening of the trace, with low amplitude fast activity ('desynchronization'), followed by synchronous activity, before any spike and wave activity develops.

As previously mentioned, considerable confusion has developed between the two quite distinct conditions of myoclonic epilepsy and infantile spasms with subnormality. These differences may be tabulated as follows.

Differences between Myoclonic Epilepsy and the Infantile Spasms Syndrome

(1) **Age of onset.** The great majority of cases of infantile spasms have their onset before the end of the first year of life. In only 1 of the 14 cases reported here did attacks start before the age of 1 year and the average age of starting myoclonic attacks was $3\frac{1}{2}$ years.

(2) **Preservation of intellect.** Almost 90% of the cases with infantile spasms develop severe

mental (and to a lesser extent, physical) retardation (Jeavons and Bower, 1964). In contrast, of the 14 children in the present series, 8 were attending a normal school and the remaining children were able, to a varying extent, to benefit from education.

(3) **EEG.** The EEG differs in the two forms of epilepsy, the children with infantile spasms showing the pattern of typical or modified hypsarrhythmia, while the children with myoclonic epilepsy show bouts of atypical spike and wave activity often with polyspikes (see Table III).

(4) **Differences in sex incidence.** In children suffering from infantile spasms, the sex incidence is 2 male to 1 female (Jeavons and Bower, 1964), whereas the 14 children reported here show an equal sex incidence which approximates to the over-all incidence of epilepsy in childhood (excluding infantile spasms), which is 1.2 male to 1 female.

(5) **Mortality.** The mortality for all cases of infantile spasms is of the order of 13% (Jeavons and Bower, 1964), while after prolonged follow-up none of the children presented here had died and indeed all enjoyed normal health between seizures.

Summary

True myoclonic epilepsy is a rare but clear-cut form of epilepsy in childhood, with its own characteristic clinical and EEG features.

Fourteen cases are described and contrasted with other forms of childhood epilepsy, in particular with the infantile spasms syndrome. Attacks consisted of a sudden, severe, and momentary muscular contraction resulting in a sudden jerk forwards or backwards of head and body and often an associated jerk of the arms. Attacks frequently caused injury by a violent fall or by striking a nearby object with head or limbs, but consciousness was seldom lost. The average age of onset was $3\frac{1}{2}$ years. In 9 children the onset of myoclonic attacks was preceded by other forms of fit.

A family history of epilepsy was unusual, and abnormal neurological signs were found in only 2 children.

Serial EEG recordings showed atypical spike and wave discharges, often with polyspikes: 3 children showed photic sensitivity.

The epilepsy was resistant to drug therapy but showed a tendency to improve spontaneously over the years.

Although some degree of mental retardation occurred in 6 children, this was not severe enough

to render them ineducable, and 8 out of 14 children were attending normal schools.

My thanks are due to Dr. B. D. Bower for suggesting a study of this group of children, most of whom were originally under his care, and for help and advice during the preparation of this paper; to Dr. P. M. Jeavons for EEG data and interpretation, as well as much helpful criticism; and to Miss K. Head and Miss J. Bruce, for obtaining serial EEG recordings on the children.

Dr. W. H. P. Cant kindly allowed me to use details of Case 13, and Dr. B. S. B. Wood of Case 7; the latter also criticized the manuscript.

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