A young boy experiencing drop attacks: what do you do next?

Commentary by

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A 4-year-old boy has been experiencing headaches and has a past history of unusual drop attacks. He has no other obvious neurological abnormality other than a mild and variable left strabismus. What are the differential diagnoses to be considered, and what are the next steps needed to establish the diagnosis?

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CASE SCENARIO

Jack, a 4-year-old boy, has recently been experiencing a few headaches and was brought in for a consultation by his mother. He was an alert and active child who was in no distress and, on examination, had no obvious neurological abnormality other than a mild and variable left strabismus (which had been present for years). His past history proved interesting; his mother said that he had been 'born with the cord around his neck', but that he did not require any prolonged intensive care and has subsequently met all his developmental milestones. From the age of 2 years, however, he has experienced unusual drop attacks. She said that when Jack was excited and running around, he would suddenly stop rigid and then fall backwards. By the time an adult had run over to pick him up, he had recovered and seemed otherwise normal. There were no postictal phenomena.

His mother initially thought Jack was acting, but after some time realised that he was not doing it voluntarily. Over the past couple of years Jack has experienced seven or eight of these episodes – all identical – the most recent of which had not been associated with his excitability. Jack had been referred six months previously to a neurologist and had undergone an EEG. His mother had been informed that he was not having seizures.

What could be happening to Jack?

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COMMENTARY

Differential diagnoses

Jack presents with a history of infrequent paroxysmal 'drop attacks' and new-onset headaches. He has a normal neurological examination. Differential diagnoses that should be considered include:

- space-occupying lesion
- seizures (tonic, atonic, myoclonic)
- movement disorder (paroxysmal kinesigenic dyskinesia [PKD], hereditary episodic ataxia type 1)
- sleep disorder (cataplexy).

Space-occupying lesion

Despite the normal neurological examination, a space-occupying lesion cannot be excluded. It is important to characterise the

nature of the headaches and the strabismus. Although the description of the strabismus (i.e. variable) suggests it is an intermittent esotropia, it is necessary to rule out a paralytic strabismus, especially a convergent strabismus due to a sixth nerve palsy. The presence of a headache in a young child requires neuroimaging. Sinister headaches (those that awaken the patient from sleep or are associated with vomiting) or a paralytic strabismus would make cranial imaging an urgency. Posterior fossa tumours, and rarely colloid cysts of the third ventricle, may present with sudden loss of consciousness, diplopia or chronic headaches.

Seizures

Although reassuring, an EEG that did not capture the episode being investigated has only limited value in ruling out seizures. Tonic, myoclonic or atonic seizures may present as 'drops'. However, the association with excitement and exercise, and the lack of postictal drowsiness do not favour a seizure. Capturing an episode during the EEG would be diagnostic, but a challenge when the episodes are infrequent and not easily provoked. A prolonged ECG recording and sleep deprivation prior to the recording may improve the possibility of recording epileptiform activity.

Movement disorder

PKD presents during childhood or adolescence and is characterised by a movement disorder (dystonia or choreoathetosis) triggered by sudden voluntary movements. If severe, the attacks may cause the patient to fall. The episodes are brief (lasting less than 30 seconds) but often occur more frequently than in Jack's case (up to 100 episodes a day). The diagnosis of paroxysmal kinesigenic dystonia is established by closely observing an episode, identifying that it is triggered by sudden movement and is associated with an abnormal posturing. PKD is usually very responsive to low-dose phenytoin, carbamazepine or sodium valproate (all used off label).

Hereditary episodic ataxia type 1 (EA-1) is characterised by brief episodes (lasting up to two minutes) of ataxia, and may be triggered by excitement, exercise or stress. Dystonic posturing may also be seen during the episode. Onset is in childhood or early adolescence. Neuromyotinia and myokymia may be seen clinically and on electromyogram between episodes. The diagnosis may be confirmed on molecular genetic testing (mutations in the *KCNA1* gene). Acetazolamide or phenytoin (both used off label) is effective treatment in some, but not all, cases of EA-1.

Sleep disorder

Cataplexy is characterised by a sudden loss of tone, from wakefulness. Episodes may last for only 30 seconds and are often triggered by laughter or anger. Cataplexy is commonly associated with narcolepsy (periods of extreme drowsiness and disturbed sleep rhythm) and sleep paralysis (inability to move for a short period of time on awakening), although young children may not report the latter. A diagnosis of cataplexy is established by performing a sleep study and a multiple sleep latency test (MSLT).

Establishing a diagnosis

In Jack's case, to establish a diagnosis it is important to:

- take a detailed history that includes a description of the headaches and of the episodes, including triggering factors, duration, associated abnormal movements and responsiveness following the episode. If Jack's mother is able to video an episode, then review of this record would be very helpful. It would also be necessary to know if there had been a change to his sleep rhythm and if he had episodes of extreme drowsiness
- perform a detailed neurological examination and look for any evidence of increased intracranial pressure (papilloedema) or paralytic strabismus
- perform an ECG and a cardiac echocardiogram because these would be useful in excluding a cardiac cause in this syncope-like presentation
- refer Jack for neuroimaging. Except in an urgent setting, where a CT may be easier to obtain and perform, an MRI is the optimum method of neuroimaging because it provides superior resolution, especially when looking for subtle cortical lesions that may be associated with seizures. There are also concerns about the radiation associated with a CT scan. However, an MRI would require a general anaesthetic in a patient of Jack's age
- attempt to observe an episode to determine if it resembles any of the differential diagnoses listed above. This may be achieved by observing Jack in an in-hospital setting
- consider a prolonged EEG recording. Although capturing an episode while the EEG is running is most useful, interictal epileptiform abnormalities may also point towards the diagnosis. Rapid transition to sleep rhythms on the EEG and loss of muscle artefact on the attached EMG leads suggest narcolepsy and cataplexy
- consider referral to a sleep physician to perform a sleep study and MSLT.

Summary

Although infrequent, Jack's episodes occur without warning and are possibly life-threatening. As the diagnostic possibilities listed above are potentially treatable, it is important to make a concerted effort to characterise an episode and intensively investigate the cause. The treatment options in Jack's case depend on the results of these investigations, and the condition the episode most resembles. MT

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