The child with funny movements
Case 2

Dr Gabriel Dabscheck

Case 2

A 12-year-old boy presents with 3 months of episodic arm movements. He has no significant past medical history.

When he stands up, or starts to run, his left hand curls up and his face contorts, for 20 - 40 seconds.

He has preserved consciousness. The events happen when there is no one watching, as well.

What is the diagnosis ?

- A. Focal seizure, preserved awareness.
- B. Movement disorder
- C. Functional neurological disorder
- D. Tic
- E. None of the above

What is the diagnosis ?

- A. Focal seizure, preserved awareness.
- B. Movement disorder
- C. Functional neurological disorder
- D. Tic
- E. None of the above

Examination

The examination was normal.

What additional tests do you do in clinic ?

- A. Hyperventilate
- B. Run
- C. Squats
- D. None of the above

What additional tests do you do in clinic ?

- A. Hyperventilate
- B. Run
- C. Squats
- D. None of the above

Additional History

The patient’s dad tells you that the elder brother presented four years ago with a similar presentation and the EEG was normal and he was told he had conversion disorder.

Psychology did not help him.

He continues to have the movements on running and standing quickly. He stopped playing sport.

Paroxysmal Kinesigenic Dyskinesia

- Rare – but not too rare (no good data)
- Clinically- sudden attacks of unilateral or bilateral movements (can include dystonia, chorea, ballism, or athetosis.
  - Onset between ages one and 20 years
  - A kinesigenic trigger (e.g., sudden voluntary movements or being startled)
  - An aura preceding attacks (10% of individuals)
  - Short duration (typically <1 minute)
  - High frequency (sometimes as many as 100 times/day)
  - No loss of consciousness or pain during attacks
- EEG, MRI, Exam – NAD
- How to treat?

https://www.ncbi.nlm.nih.gov/books/NBK475803/

What medication do you prescribe?

- A. Levetiracetam
- B. Carbamazepine
- C. Amitriptyline
- D. Oxcarbazepine
- E. None

What medication do you prescribe?

- A. Levetiracetam
- B. Carbamazepine
- C. Amitriptyline
- D. Oxcarbazepine
- E. None

Paroxysmal Kinesigenic Dyskinesia

- Autosomal dominant condition (10% de novo)
- Mutations to PRRT 2 (also gene implicated in benign familial infantile epilepsy and hemiplegic migraine) – no good genotype phenotype correlation exists. 60 – 90 % penetrant.
- Prevention or control with phenytoin or carbamazepine
  - Low dose Carbamazepine or Oxcarbazepine are very effective.

Take Home Message

- Action induced movement disorders – think of PKD
- It is treatable