

A case of episodic paralysis

A 43 year old man presented with episodes of limb weakness since childhood.

HISTORY

At the age of 9, the patient experienced his first episode of paralysis, which was initially thought to be due to poliomyelitis. At this time he was unable to move his arms and legs, but was able to talk and swallow and could open his eyes. It had been preceded by a non-specific 'flu-like illness two weeks before the ictus. The illness and paralysis lasted about one week and he made a full recovery. His second major episode occurred at the age of 19. Since then he has had rather frequent attacks. In his 20s and 30s, he seemed to have about one per year of varying severity. He has had a maximum of 3 per year.

A typical attack will come on acutely and affect arms, legs or both. At their worst he has complete paralysis of his whole body including facial muscles. He is able to talk and swallow and remains fully conscious throughout an attack. He has never needed ventilation on an intensive care unit. A typical attack lasts for 24-48 hours. He also has had more minor episodes with short-lived (24-48 hours) periods of severe arm weakness. Resolution is spontaneous and full. There is no history suggestive of episodes of ataxia. In between attacks he has no significant problems with his limbs. There is no longstanding weakness or myalgia.

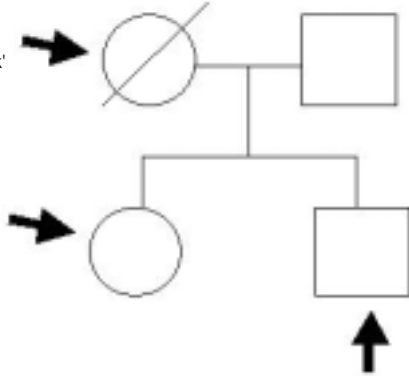
In addition to these symptoms, he had features suggestive of Raynauds, with severe pain in his fingers in the cold.

His PMH is unremarkable and he has no cardiac problems.

The Family History is interesting. His mother died at the age of 34 from an apparent MI and she too had episodes of weakness. His sister is under the care of neurologists with similar problems.

Episodic weakness
Died @ 35
'heart attack'

Episodic weakness



He took no medications and worked full time. He did not drink or smoke.

EXAMINATION

His general examination, including cardiovascular exam was for the most part unremarkable. He had mild hypertelorism, low set ears and mild micrognathia. The hands showed evidence of clinodactyly. There were no neurological signs, specifically no limb weakness or cerebellar signs.

INVESTIGATIONS

He had over the last 10 years been extensively investigated, with the results given below:

- Routine bloods: Normal. No acute abnormalities documented during an attack, specifically serum potassium levels normal.
- Autoantibody screen: negative
- EMG/NCS: normal, with no myopathic or myotonic features
- Muscle biopsy: 'very mild non-specific myopathic changes. No evidence of fibre vacuolation'.
- ECG and 24-ECG tape: normal

TREATMENTS

He had previously tried amiloride, K supplements and acetazolamide with little apparent effect. He is currently on no medication.

DIAGNOSIS

His sister who is under the care of neurologists has been found to have a mutation in her *KCNJ2* gene which encodes the Kir2.1 ion channel. Thus the diagnosis is Andersen's syndrome.



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