SEIZURES OR BEHAVIOR DISTURBANCES?

A case report on Cornelia de Lange Syndrome

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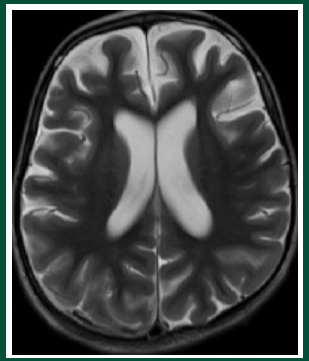
THE CASE

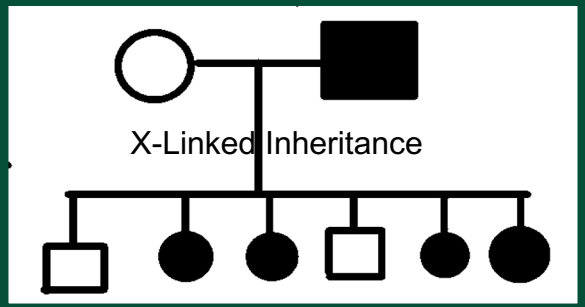


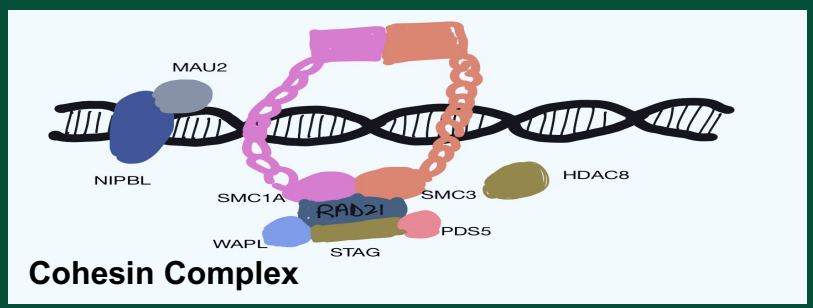
- 20-year-old male with a minor phenotype of Cornelia de Lange syndrome (IQ 50-60).
- Exacerbation of episodic behavioral disturbances- sub acutely started a year ago, worsening
- Recently discharged from inpatient psychiatric unit after a 30 day stay- on Citalopram & Oxcarbazepine
- Previously tried multiple antipsychotics which led to worsening of symptoms. Vitals & Labs -WNL
- Difficulties in placement- treated in the emergency department. CT- negative for acute Process
- Patient did not tolerate EEG. Behaviors followed a specific sequence.
- Responded initially to low dose Lorazepam. Oxcarbazepine titrated to complete resolution of symptoms. Total stay - 5 days.

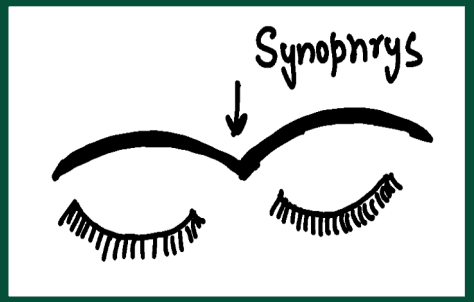
Flinging movement of left arm → Bilateral swinging of upper arms → Sharp Cry → Staring in Space- few minutes → Agitation

DISCUSSION

Cornelia de Lange Syndrome is a rare genetic disorder with an incidence of 1 in 10,000. Consequence of a mutation in cohesin-associated genes(NIPBL, SMC1A, SMC3 , RAD21, HDAC8). SMC1 mutations are responsible for about 5% of the case. It follows X-linked, familial inheritance. Mutations are fully penetrant, affect males and females equally. Dysmorphic features include - short stature, hypertelorism,synophrys, thin down turning upper lip. Seizures are most associated with SMC1 mutation. Partial epilepsy is the most common epilepsy associated with the syndrome with focus in temporal and parietal areas. ,The patient's poor response to antipsychotics, repetitive patterning in patient's symptoms and response to antiepileptics increase suspicion of a seizure disorder







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HARRISON MOYNIHAN MS4 DO CANDIDATE

References available upon request