A rare cause of ovarian acquired failure: ovarioleucodystrophy

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Background: Ovarianleucodystrophies are one of the rarest leucodystrophies associated with primary ovarian failure. Patients may present with variable disease manifestations such as neurologic, psychiatric or ovarian failure. Disease onset may occur in infancy, adolescence or adulthood caused by mutation in the eukaryotic initiation factor 2B (eIF2B) which has a poor prognosis.

Objective and hypotheses: Seventeen-year-old girl was brought with tremor, gait difficulty, dysarthria and tingling. She was born at term with a birth weight of 2500 g. and her parents were cousins. It was learned that telarche started when she was 12 years old and was evaluated for primary amenorrhea when she was 16 years old. Her chronological age was 17 years, height 162 cm (46 p), weight 51.5 kg (35 p), and puberty was consistent with Tanner stage 5. She had no signs of hyperandrogenism and it was thought that she had premature ovarian failure with high gonadotropin levels. FSH: 112 U/l, LH: 42 U/l, estradiol: <11 pg/ml, thyroid functions, prolactin, α-fetoprotein ve β-HCG were in normal ranges. Uterus sizes were 50×15×17 mm with bilateral atrophic ovaries and karyotype was 46,XX. Brain MRI tested for neurologic symptoms was compatible with leucodystrophy. Ovarian failure and leucodystrophy key words leaded us to conclusion.

Conclusion: A global view prevents to be drown in details. Symptoms should be assessed totally instead of separately. This autosomal recessive entity should be kept in mind before further investigation.