

When height hides a mitochondrial mystery: Primary amenorrhoea beyond gonadal dysgenesis

*Akther F¹ , Huq A² , Tofail T³ , Banu H⁴ , Sultana N⁵ , Hasanat MA⁶ 

¹Fahamida Akther, Phase B resident, Department of Endocrinology, BMU, Dhaka, Bangladesh; ²Afrin Huq, FCPS Trainee, Department of Endocrinology, BMU, Dhaka, Bangladesh; ³Tania Tofail, Assistant Professor, Department of Endocrinology, BMU, Dhaka, Bangladesh; ⁴Hurjahan Banu, Associate Professor, Department of Endocrinology, BMU, Dhaka, Bangladesh; ⁵Nusrat Sultana, Associate Professor, Department of Endocrinology, BMU, Dhaka, Bangladesh; ⁶MA Hasanat, Professor, Department of Endocrinology, BMU, Dhaka, Bangladesh

Abstract

Background: Primary amenorrhea with tall stature is an unusual combination that, when accompanied by neurological features, warrants evaluation for syndromic and mitochondrial causes beyond classical gonadal dysgenesis syndromes.

Case summary: A 21-year-old girl presented with primary amenorrhea, absent breast development and progressive height gain. Her medical history was notable for recurrent seizures since infancy, subsequently evolving into epilepsy, along with type 2 diabetes mellitus, strabismus with diplopia and frequent falls. She was born to consanguineous parents with a significant history of adverse pregnancy outcomes, including a sibling who died following convulsions. Examination revealed tall stature (178 cm; SDS +2.9) with eunuchoid body proportions (upper-to-lower segment ratio, 0.8; arm span, 200 cm), Tanner stage B1P3A3, and normal female external genitalia without virilization. Hormonal evaluation demonstrated hypergonadotropic hypogonadism. Karyotyping revealed a 46, XX pattern. Pelvic ultrasonography revealed a hypoplastic uterus and streak-like ovaries. Bone age was delayed. Brain MRI demonstrated periventricular leukomalacia with corpus callosum defect. Combined endocrine, metabolic and characteristic neuro-ophthalmic findings are highly suggestive of an AARS2-related disorder. However, molecular confirmation is yet to come.

Conclusion: This case highlights AARS2-related disorder as an important, under-recognized cause of primary ovarian insufficiency, with tall stature when accompanied by seizures and oculomotor abnormalities and underscores the value of genetic testing in unexplained combined neuroendocrine presentations, particularly in consanguineous families. [*J Assoc Clin Endocrinol Diabetol Bangladesh*, 2026;5(Suppl 1): S54]

Keywords: AARS2-related disorder, Primary amenorrhea, Primary ovarian insufficiency, Tall stature, Hypergonadotropic hypogonadism, Diabetes mellitus, Epilepsy

***Presenting & Corresponding Author:** Dr. Fahamida Akther, Phase B resident, Department of Endocrinology, BMU, Dhaka, Bangladesh, E-mail: fahamida41st@gmail.com, ORCID iD: <https://orcid.org/0009-0008-0401-6831>