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Tetrasomy X (48,XXXX): A Rare Chromosomal Aneuploidy with Reproductive Impact

*Hossain SM[1], Islam HM[2], Banu H[3], Sultana N4, Hasan M[5], Morshed SM[6], Tofail T[7], Jahan S[8], Hasanat MA[9]

[1]Md Saddam Hossain, Resident, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [2]Md Hasanul Islam, Resident, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [3]Hurjahan Banu, PhD Research Fellows, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [4]Nusrast Sultana, PhD Research Fellows, Department of Endocrinology, Bangladesh Medical University, Dhaka; [5]Mashfiqul Hasan, PhD Research Fellows, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [6]Md Shahed Morshed, PhD Research Fellows, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [7]Tnia Tofail, PhD Research Fellows, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [8]Sharmin Jahan, Professor, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh; [9]M A Hasanat, Professor, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh

Abstract

Tetrasomy X (48,XXXX) is an exceptionally rare sex chromosome aneuploidy, with fewer than a few hundred cases documented worldwide. Clinical features vary widely, from mild developmental delay to pronounced dysmorphism, gonadal dysfunction, and tall stature. We report a young female who presented with menstrual irregularities and delayed pubertal development. On physical examination, she was unusually tall (height 182 cm, arm span 194 cm) with a low upper-to-lower segment ratio (0.91), consistent with eunuchoid body proportions. Tanner staging revealed discordant pubertal progression, with breast development at stage B4 but pubic hair at stage P1. Laboratory evaluation demonstrated hypergonadotropic hypogonadism, evidenced by elevated follicle-stimulating hormone (37.93 mIU/ml), elevated luteinizing hormone (16.61 mIU/ml), and low estradiol (17.21 pg/ml). Other biochemical and systemic evaluations, including thyroid, renal, hepatic, and cardiac assessments, were within normal limits. Karyotyping confirmed 48, XXXX, establishing the diagnosis of tetrasomy X. This case emphasizes the diagnostic challenge posed by such rare aneuploidies, particularly in individuals presenting with tall stature and reproductive dysfunction but without overt dysmorphic features. Early recognition is crucial for initiating hormonal replacement, addressing fertility concerns, and ensuring multidisciplinary long-term care. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S60]*

Keywords: Tetrasomy X, 48,XXXX, Chromosomal aneuploidy, Hypergonadotropic hypogonadism ,Delayed puberty, Tall stature

Presenting & Corresponding Author: Dr. Md Saddam Hossain, Resident, Department of Endocrinology, Bangladesh Medical University, Dhaka, Bangladesh**.** Phone: +8801763322621, email: saddamimc6@gmail.com