P96

Isochromosome Xq in Mosaic Turner Syndrome: A Case Report

Dr Mark Ramon Victor Llanes[1], Dr May Naranjo[1]
[1]*Southern Philippines Medical Center, Davao City, Philippines*

Isochromosome Mosaic Turner Syndrome (IMTS) is a variant of Turner Syndrome (TS) characterized by cytogenetic profile of 1 or more additional cell lineages aside from 45,X, and the presence of a structurally abnormal X chromosome consisting of either two short or two long arms. IMTS is rare, with only 8-9% prevalence among women with TS based on international studies, and 15% of all TS in the Philippines.

A 20 year old female came in due to amenorrhea and alopecia. Physical examination revealed short stature, cubitus valgus and Tanner Stage 1 pubic hair and breast development.

Transrectal ultrasound revealed absent ovaries and infantile uterus. Hormonal evaluation revealed hypergonadotropic hypogonadism. Bone aging was that of a 13 year old for females with non fusion of epiphyseal plates. Cytogenetic study revealed 45,X [37]/46, X, i (X) (q10) [13]. This is consistent with a variant Isochromosome Mosaic Turner Syndrome.

She was screened for medical complications. Audiogram and two-dimensional echocardiography were unremarkable. She has dyslipidemia and was given statins. She has subclinical hypothyroidism with positive test for anti-thyroglobulin antibody. Her intelligence quotient (IQ) was below average. She received conjugated estrogen and progesterone that patterned the hormonal changes in normal menstrual cycle. On the third week of hormonal therapy, she developed breast mound and on the fourth week, she had her first menstrual period. Her alopecia resolved spontaneously.

The above case is a rare variant of Turner Syndrome requiring supportive, medical and psychological care.