Consultant360 Multidisciplinary Medical Information Network

CASE IN POINT

PEER REVIEWED

Turner Syndrome With Associated Behavior Disorder

Authors:

Syed A. A. Rizvi, PhD, MS, MBA

Hampton University School of Pharmacy, Hampton, Virginia

Zafar Qureshi, MD

UHI CommunityCare Clinic, Miami, Florida

Syed A. Hassan, BS

Florida International University, Miami, Florida

Hassan Arif, BS

University of South Florida, Tampa, Florida

Mohammad J. Latif-Jangda, MD

Nova Southeastern University, Fort Lauderdale, Florida

Zaib Latif-Jangda, BS

Nova Southeastern University, Fort Lauderdale, Florida

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A 14-year-old girl presented with anxiety disorder due to her short stature resulting from mosaic Turner syndrome. She also had autism, microcephaly, severe intellectual disability, and profound vitamin D deficiency.

Her mother described the girl's general mood as "sad" and stated that her daughter doesn't like many of the other kids in her class nor really interacts well with them. The girl had had no hospitalization, surgery, head trauma, or fracture, and she denied headache, blurry vision, polyuria, polydipsia,

nocturia, bedwetting, hip pain, nose bleeding or easy bruising, heart disorders, and bowel problems. She had had gum bleeding, and her mother had noted blood on the girl's pillow. She had not had her first her first menstrual period yet and previously had been on growth hormone treatment.

Physical examination. The patient was oriented to person, place, and time. She appeared well-developed and well-nourished but has short statue. The remainder of the physical examination findings were unremarkable. Her blood pressure was 98/59 mm Hg, pulse was 70 beats/min, and respiratory rate was 17 breaths/minute. The patient's height was 137.9 cm (89% based on Turner syndrome data) and weight was 30.1 kg, with a body mass index of 15.83 kg/m^2.

Diagnostic tests. Laboratory test results included the following values: red blood cell count, 5.24 × 10^6/μL; white blood cell count, 4400/μL; hemoglobin, 13.2 g/dL; sodium, 141 mEq/L; potassium, 4.1 mEq/L; calcium, 10.0 mg/dL; albumin, 4.6 g/dL; aspartate aminotransferase, 22 U/L; alanine aminotransferase, 11 U/L; creatinine, 0.51 mg/dL; blood urea nitrogen, 9 mg/dL; random glucose, 95 mg/dL; serum estradiol, 1.5 pg/mL; follicle-stimulating hormone, 101 mIU/mL; luteinizing hormone, 18 mIU/mL; C-peptide, 1.6 ng/mL; free thyroxine, 1.36 ng/dL; and 25-hydroxyvitamin D, 16.4 ng/mL (reference range, 30-100 ng/mL).

Routine chromosome genetic testing showed pericentric inversion of chromosome 9 in all cells, which is not pathogenic. She had mosaic 45,X/46,X,rX Turner syndrome as established by a 45,X karyotype in 16 of 25 (64%) cells and a 46,X,rX karyotype (denoting a ring X chromosome) in 9 of 25 (36%) cells. Ring chromosomes involve fusion of chromosome ends with deletion of terminal DNA that in this case would include the X short-arm sequences that are most crucial in suppressing Turner syndrome findings.

Discussion. Turner syndrome (after Henry H. Turner, who in 1938 first described the condition[1]), is a female-specific disorder affecting approximately 1 in 2,500 individuals[2] in which part or all of a normal second sex chromosome is missing, leading to various structural abnormalities.[3,4] In terms of distribution, approximately 50% of persons with Turner syndrome have monosomy X (45,X), some have duplication of the long arm of one X chromosome (46,X,i[Xq]), and 30% to 40% display mosaicism (45,X/46,XX) with a normal cell line.[4-6]

Approximately 2% to 11% of persons with Turner syndrome have a ring X chromosome and a mosaic 45,X/46,X,rX karyotype. These individuals generally have relatively milder symptoms compared with individuals with a nonmosaic 45,X karyotype; however, they have an increased risk (especially in the presence of a ring) of mental retardation.[4,7,8] Turner syndrome is also known to be associated with a higher risk of autism,[9] and co-occurrence of macrocephaly also has been reported.[10]

Most observed clinical features in women with Turner syndrome include webbed neck, short stature, cardiac and renal malformations, osteoporosis, hearing problems, hypertension, hypothyroidism and

cardiac and renal malformations, osteoporosis, hearing problems, hypertension, hypothyroidism and gonadal dysgenesis. Affected individuals may present with varying degrees of clinical symptom at various stages of life.[11] Approximately 70% of patients with Turner syndrome are reported to have learning disabilities, approximately 10% will have significant developmental delays, and approximately 2% to 10% will have coexisting psychiatric diagnoses.[4,12-14] Our patient's sexual immaturity and short stature are compelling reasons for her to feel anxious, sad, and isolated from peers.

Often parents of children with Turner syndrome need counseling and support due to the feeling of guilt and risk of poorer adjustment.[15,16] In our patient's case, the presence of both microcephaly and autism is very unusual in classic Turner syndrome, and the mother's reactions might be expected to exceed the usual parental concerns over mild learning differences and short stature. Our patient's mother, the sole caregiver and a single mother, completed a Caregiver Burden Scale, a 22-item questionnaire. Her score was 21, indicating that she experiences a mild to moderate burden. Of note in her responses, she reported that she "nearly always" feels stressed between caring for her daughter and trying to meet other responsibilities for her family or work, is afraid about what the future holds for her daughter, and feels that she has lost control of her life since her daughter's illness.

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