Turner Syndrome with Hypertension

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A sixteen-year-old Bahraini female with a history of uncontrolled hypertension presented with amenorrhea for which ultrasound abdomen revealed rudimentary ovaries (streak gonads) with normal sized kidneys. Laboratory and diagnostic data were all normal with a blood pressure of 180/95 mmHg. However, 24-hour ambulatory BP monitor confirmed hypertension with an average BP of 177/100 mmHg.

The patient had normal IQ and a short stature with webbed neck, low-set ears with widely-spaced nipples and increased carrying angle; all these characteristics were typical of Turner syndrome. A blood sample of the patient was sent for karyotype analysis which showed an abnormal karyotype with monosomy X, such as 45,X0[20%]. Chromosome analysis had confirmed the diagnosis of Turner syndrome which correlates with the other test results since over 50% of young females with Turner syndrome experience abnormal blood pressure similar to that of adult patients with secondary hypertension.

Bahrain Med Bull 2019; 41(3): 177 - 178

Turner syndrome (TS) is a chromosomal condition affecting 1 in 2,500 newborns in which a female lacks one or a partial sex chromosome X. Although the diagnosis of TS can be made prenatally or during infancy, girls with mild signs and symptoms are typically diagnosed late during the teen or young adult years[1]. It is common that females with this syndrome experience developmental issues such as early loss of ovarian function and/or amenorrhea.

Females with TS usually have a webbed neck, skeletal abnormalities and increased carrying angle and most girls will have normal intelligence. One-third to one-half of Turner syndrome patients have kidney problems or are born with a cardiovascular defect such as coarctation of the aorta and/or high blood pressure[2]. Arterial hypertension is present in approximately 13-58% of adults with TS and in approximately 25% of pediatric patients[3].

The aim of this presentation is to report a case of a female with Turner syndrome suffering from uncontrolled hypertension.

THE CASE

A sixteen-year-old Bahraini female with a history of uncontrolled hypertension presented with amenorrhea. Ultrasound abdomen revealed rudimentary ovaries (streak gonads) with normal sized kidneys but a blood pressure of 180/95 mmHg.

The patient had normal IQ, achieving high grades at school. Due to her young age, secondary hypertension was excluded. She weighs 57 kg with a height of 141 cm. Physical examination revealed short stature with webbed neck, low set ears with widely spaced nipples and increased carrying angle was exhibited, all characteristics were typical of Turner syndrome.

She was prescribed Coveram 5mg tablet once daily which is a combination of perindopril 5 mg and Amlodipine 5 mg but her blood pressure was uncontrolled and she suffered from a cough induced by angiotensin-converting enzyme (ACE) inhibitor. Coveram was stopped and a 24 hours ambulatory blood pressure monitor had confirmed hypertension with an average blood pressure of 177/100 mmHg.

Electrocardiogram revealed normal sinus rhythm and showed a regular heart rate of 85/minute. Her CT aortogram was normal, which excluded coarctation of the aorta. 2D echocardiography and chest X-ray were also normal, her kidneys had a normal function with a serum creatinine value of 48 μmol/L, estimated GFR was 95 mL/min per 1.73 m^2, 24-hours urinary protein was 0.2 g/d and a Renal Artery Duplex scan showed no evidence of renal artery stenosis.

Laboratory tests were all normal with a TSH value of 1.8 mU/L, blood sugar of 3.8 mg/dl; potassium was 4.8 mEq/L and unremarkable lipid profile.

Peripheral blood lymphocytes were cultured for 72 hours using PBMAX media. Cells were harvested and banded with Giemsa (GTG-banding). Twenty cells were captured and analyzed at a resolution of 400 using the Leica CytoVision software. The karyotype of the patient can be seen in figure 1.

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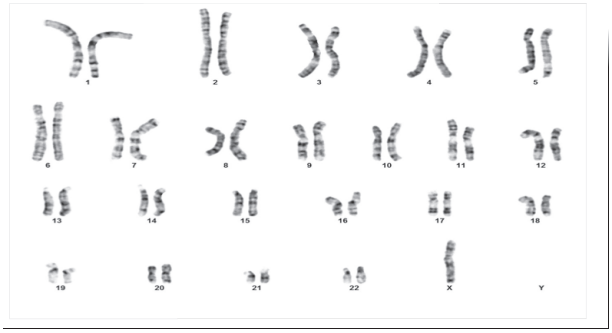

Figure 1: Karyotype of GTG-banded chromosomes at a resolution of 400 revealed monosomy X 45,X0[20%]

DISCUSSION

The diagnosis of Turner syndrome would explain the high blood pressure, since 40% of patients have parenchymal renal disease that leads to hypertension but other causes of hypertension in Turner syndrome like coarctation of the aorta, horseshoe kidney or renal artery stenosis were excluded.

An abnormal karyotype was visualized for the patient with monosomy of chromosome X 45, X0[20%]. There were no other numerical/structural chromosomal abnormalities seen. Hence, chromosome analysis had confirmed the diagnosis of Turner syndrome which correlates with the other test results since over 50% of girls with Turner syndrome experience abnormal blood pressure similar to that of adult patients with secondary hypertension[4].

Many studies revealed the association of hypertension with Turner syndrome. Most studies reported higher BP values in TS patients compared to healthy controls of similar age[4]. Hypertension was also found to be more common in patients with TS than in the general population[5]. In another study, women with TS were more likely to be hypertensive than other females and the hypertension is independent of obesity[6].

Insufficient information can be found regarding the cause of hypertension in Turner syndrome because it is poorly understood and presumably multifactorial[3]. Potential factors involved in the pathogenesis of hypertension include inappropriate activation of the renin-angiotensin-aldosterone (RAA) system, oxidative stress, inflammation, impaired insulin-mediated vasodilatation, increased stimulation of sympathetic nervous system and abnormal sodium processing by the kidney[7].

Upon prescription of Coveram, our patient had suffered from a cough in response to ACE enzyme; a similar outcome was seen in one study[8]. It is possible that patients with TS, experience overactivation of the sympathetic nervous system which increases the blood pressure and heart rate[3]. It is possible that there are certain critical genes on the second X chromosome that are defective in those living with TS and it is the loss of these genes that trigger such an effect[9].

The patients' blood pressure should be monitored regularly. A healthy lifestyle must be adopted and that includes having a nutritious diet, enough sleep and physical activity.

CONCLUSION

Cytogenetic evaluation in patients who have clinical features resembling Turner syndrome is important. Although the cause of hypertension in patients with Turner syndrome is still unclear, their association has been clinically and statistically confirmed. Management and treatment of patients with Turner syndrome demand a multidisciplinary approach.

Author Contribution: All authors share equal effort contribution towards (1) substantial contributions to conception and design, acquisition, analysis and interpretation of data; (2) drafting the article and revising it critically for important intellectual content; and (3) final approval of the manuscript version to be published. Yes.

Potential Conflicts of Interest: None.

Competing Interest: None.

Sponsorship: None.

Acceptance Date: 27 May 2019.

Ethical Approval: Approved by the Research and Ethics Committee, King Hamad University Hospital, Bahrain.

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