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Case Study

Adult Turner Syndrome – A Family Physician's Perspective

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ABSTRACT

This patient had delayed puberty and was diagnosed as having Turner syndrome in adulthood. There are multiple issues to be addressed, not just the medical conditions and the ovarian failure but also the fertility options, the psychosocial aspects and the religious issues that present with the condition. As a family physician, many important issues need to be addressed, taking a holistic approach to ensure comprehensiveness and continuity of care.

Keywords: Turner syndrome, delayed puberty, fertility options, psychosocial issues

INTRODUCTION

Turner syndrome (TS) was first described by an anatomist in 1768 but was named after Dr. Henry Turner in 1938, an endocrinologist who had a series of patients with characteristics of the syndrome.[1] Turner syndrome is a chromosomal condition affecting females of unknown aetiology. It is not hereditary and occurs 1 in 2500 live born girls. The clinical features vary. The diagnosis is usually made early in childhood due to characteristic features or in adolescence due to delayed puberty.[2]

THE CASE

Miss SZ, a 23-year-old single Muslim lady presented to a government health centre with a complaint of amenorrhoea. On further questioning, it was found that she had never attained menarche and her secondary sexual characteristic were underdeveloped. There was no significant past history nor family history. Miss SZ is the 6th child of nine siblings. Both her parents are in their 50s and it was a non-consanguineous marriage. All her siblings are male. She studied until form five in Perak and achieved third grade in her Sijil Pelajaran Malaysia (SPM) examinations. On completion of her SPM, she came to Kuala Lumpur and stayed with her cousins. She is working as a production operator in a factory in Petaling Jaya. She is not sexually active but is currently in a relationship. She presented late, only after being prompted by her cousin who told her that she is "not normal".

On examination, Miss SZ was found to be a pleasant lady. Her BMI was 26 kg/m^2 (height 149 cm, weight 58 kg). Her blood pressure was 110/60 mmHg and pulse rate was 80/min (regular) with no radiofemoral delay. There was no goitre. Her breast was infantile

(Tanner stage 1) with widely spaced nipples and a "shield-like chest". She had sparse pubic and axillary hair (Tanner stage 2). She had slight cubitus valgus. Her genitalia was normal with an intact hymen. Her cardiovascular and respiratory examinations were unremarkable. The abdomen was soft and not organomegaly. An ultrasonography of the pelvis was done which revealed a small uterus 4.2 x 0.6 x 1.7 cm; ovaries were not visualised, and no other adnexal mass was noted.

Initial investigations were done at the health centre. She was given an appointment to review the results. On the subsequent visit, her results were reviewed: Thyroid function tests: free T4 – 20.2 pmol/L (reference range: 11.5 – 23.2); TSH – 1.26uI/ml (reference range: 0.40 – 5.50). Gonadal hormones: FSH – 47.6 miU/ml (reference range: depends on cycle); LH – 18.0 miU/ml (reference range: depends on cycle); Oestradiol – < 3 (reference range: 0 – 198); Testosterone – 1.7 nmol/L (reference range: 0.5 – 2.6).

A provisional diagnosis of Turner syndrome was made and Miss SZ was informed of the diagnosis. She was counselled and further follow up was arranged to the health centre. She was also referred for shared care with a gynaecologist. During the consultation with the gynaecologist, karyotyping was done and revealed 45XO/46XX, which confirmed Mosaic Turner syndrome. She was commenced with hormone replacement therapy by the gynaecologist. A computer tomography (CT) scan of the abdomen and pelvis was arranged to confirm the absence of gonads; echocardiography and a bone mineral density study were also arranged. She was also counselled by the gynaecologist on infertility. After her appointment with the gynaecologist, Miss SZ came to the health clinic as she wanted to clarify a few concerns which she could not do in view of the given long duration follow-up with the gynaecologist.

Her concerns were related to whether she could conceive and why more investigations were arranged. With treatment she expressed the hope that she would lead a life as a normal woman. During this visit, her understanding of her condition was explored. As she had been overwhelmed with the information provided earlier by the gynaecologist, she wanted to to clarify certain issues. Her concerns and expectations in regard to being a normal woman were discussed. Explanation on why further evaluation was required and its importance was also explained to Miss SZ. After the consultation she had a better understanding of her condition. She was advised to go for regular follow-up to monitor for potential complications, both physically and emotionally. Miss SZ is now under shared care between the gynaecologist and the family physician.

DISCUSSION

This patient scenario illustrated a case of adult Turner syndrome with delayed puberty and a possible fertility problem being seen and managed in the primary care setting. Turner syndrome(TS) is usually diagnosed in childhood or when presented with delayed puberty. The diagnosis of Turner syndrome may be delayed until adulthood in 10% of women if they enter puberty spontaneously. The diagnosis is confirmed by chromosomal analysis by tissue karyotyping.[1] It is to be noted that more than 50 % of Turner syndrome cases have mosaic chromosomal complement such as 45X/46 XX. Advanced maternal age is not

associated with increased incidence of TS. The clinical manifestations in adulthood may present as short stature. The aetiology of growth failure is unknown. However the possibility of primary bone defect and partial growth hormone insensitivity could not be excluded.[1,2] They may also present with bony abnormalities such as cubitus valgus, abnormal upper to lower body segment ratio and scoliosis. Fortunately, these deformities do not cause any disability to the women with TS.[3] As a result of gonadal failure, women with TS exhibit underdeveloped sexual characteristics and primary amenorrhoea, as in Miss SZ.

There is increasing evidence that as an adult with TS, these women are more susceptible to a range of medical disorders which require long term follow-up for early intervention to reduce morbidity and mortality, hence improving quality of life.[1,2] The morbidities associated with adult TS includes osteoporosis, cardiovascular disease (up to 40% have left-sided congenital cardiac anomalies and 10% have coarctation of the aorta). Hypertension is common. The risk increases by three-fold, even in the absence of cardiac or renal disease. Individuals with TS have increased incidence of primary hypothyroidism with increasing age, and up to 50% have antithyroid antibodies. However, they do not present with overt clinical symptoms. Renal dygenesis is more common compared to the normal population.[2]

Miss SZ, had to undergo a battery of investigations. These evaluations were necessary for proper management and care to be instituted for her. In view of the risk of a number of medical problems, she requires care throughout her life. As with other chronic illnesses, continuity of care is crucial for her future care, which has to be individualised. This can be done effectively at the primary care level by a family physician as the care is usually personalised, and in this case as she had been seen three times, rapport had been established with the same physician. Furthermore, a lower consultation cost and closer vicinity to the health centre in the community provides for easy accessibility of the family physician, more so when the patient has any doubts to clarify pertaining to her condition. In this scenario, the family physician's role was not only to coordinate the care of the patient by making necessary and appropriate referral but also to provide comprehensive care and emotional support to the patient.

Individuals with Turner syndrome may have lower self-esteem and lower self-image due to their short stature and delayed sexual maturation, especially during adolescence and adulthood. Therefore, they are at increased risk of social and emotional difficulties.[2,4] At the primary care level, family physicians have an advantage as they are able to provide counselling on the issues relating to infertility and sexual relationships. With continuity of care and the establishment of a good rapport, the patient will be more receptive towards the physicians in confiding and discussing any related psychosocial issues.

In TS, more than 90% of the women have premature ovarian failure, hence, subsequent oestrogen deficiency. Therefore, they do not enter spontaneous puberty. In view of significant risk of physical and psychological morbidity related with oestrogen deficiency, oestrogen replacement plays an important role in the management of TS.[4] The aim of oestrogen replacement is to initiate puberty and to prevent morbidity related to oestrogen deficiency, such as osteoporosis.[5]

Routine counselling for sexually transmitted illness prevention is recommended as with any other females who are sexually active. The majority would require long term replacement and controversies arise on the benefits versus risks of oestrogen replacement

therapy. It has been recommended that only natural oestrogen should be used in the physiological doses to reduce the risk of breast carcinoma and progestogen should be prescribed in combination to prevent endometrial carcinoma.[3] However, recommendations for close monitoring including breast examination and mammography are similar for the general population. As part of holistic care, a family physician should also provide preventive care for the patient in terms of counseling on safe sex practices and providing screening for breast carcinoma since Miss SZ is on hormonal replacement therapy.

With regard to fertility, at present, pregnancy can be achieved by oocyte or embryo donation and in vitro fertilisation. However, these women require adequate hormonal replacement before transfer of oocyte or embryo can be done. The risk of miscarriages is also high. [1,4] According to Islamic law, oocyte donation and in vitro fertilisation are only permissible if the tissue obtained is of the patient's own and her spouse; not from another individual.[6] In Malaysia, services for oocyte or embryo donation is unavailable or rather limited. Therefore, Miss SZ was counseled on possibility of permanent infertility by the gynaecologist. It is important that the regular family physician follow-up on Miss SZ is able to provide appropriate emotional support as she may encounter emotional difficulties if her partner rejects her and she is also at risk of depression in view of her current infertile situation.

As a doctor in the community, the family physician has a unique position to deal with all the health issues faced by Miss SZ. She will require long term follow up in view of the possible complications that could arise due to Turner syndrome. The family physician will be able to identify her needs and coordinate her care appropriately including the social network support.

CONCLUSION

In this patient's scenario, it is crucial that she understands all the issues pertaining to her condition and its consequences, the significance and importance of the investigations performed and the treatment instituted. As a family physician, treating the patient comprehensively would allow a better therapeutic alliance to be established and continuity of care to be assured.

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