Sexual health needs of a patient with mosaic Turner’s syndrome

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Case report
A 26-year-old woman, who had previously been diagnosed in childhood with mosaic Turner’s syndrome, presented in general practice. The patient had a history of drug dependency and had recently undergone rehabilitation. On presentation she was not being treated for Turner’s syndrome, and it appeared that she had had no estrogen replacement therapy for several years. She had been sexually active during this time but had never used any form of contraception, as she understood that she was infertile. She had never had a cervical smear test.

A review of the patient’s records showed that she had been referred to a paediatric growth clinic aged 10 years because of short stature, although her height was found to lie between the 3rd and 10th centiles. It was initially thought that her short stature was constitutional, as she did not exhibit the clinical features of Turner’s syndrome, but chromosome analysis showed mosaic Turner’s syndrome. Some 90% of the cells were XO with the remaining 10% being XX, with the second X appearing as a ring chromosome. Her bone age was delayed by approximately 2 years. Pelvic ultrasound examination revealed no evidence of a uterus or ovaries but this examination did not appear to have been repeated as the patient matured. She was treated initially with subcutaneous growth hormone, with the addition of ethinylestradiol (EE) when aged 11 years, and the introduction of cyclical norethisterone when she developed vaginal bleeding. She had not menstruated spontaneously when oral EE was commenced. Growth hormone was stopped at age 17 years when her height was 155 cm (9th centile). Her care was then transferred to an endocrinology clinic. It does not appear from her records that she ever attended this clinic.

As part of her initial assessment in primary care the patient underwent a pelvic examination and a cervical smear was taken. The cervix appeared small but inflamed and there was clinical evidence of vaginitis. The smear showed probable Trichomonas vaginalis infection. Further testing showed Chlamydia trachomatis infection of the endocervix. Both of these infections were treated successfully. As part of the patient’s further management, including partner notification. Other sexually transmitted infection (STI) investigations proved negative and she was started on a course of hepatitis B immunisations. She was also referred to gynaecology outpatients for further assessment of her Turner’s syndrome. Blood tests subsequently revealed her hormone levels to be as follows: luteinising hormone (LH) 50.7 IU/l, follicle-stimulating hormone (FSH) 105.5 IU/l and 17 β-estradiol <73 pmol/l, all of which were in keeping with a diagnosis of ovarian failure. Pelvic ultrasound examination showed a small postpubertal uterus but no ovaries could be demonstrated. The patient was commenced on the combined oral contraceptive (COC) pill to provide her with hormone replacement therapy (HRT) and protection against the small risk of unplanned pregnancy. She also received advice regarding safer sex. Her smear test was repeated at the appropriate time and was negative. Arrangements were made for a pelvic magnetic resonance imaging scan, and an echocardiogram, but unfortunately the patient’s drug dependency syndrome relapsed and she was lost to outpatient follow-up.

Discussion
Turner’s syndrome (or Ullrich-Turner syndrome), which was first described by Ullrich in 1930 and Turner in 1938, is a relatively common chromosomal disorder occurring in approximately 1 in 2000 live births, and is the most commonly occurring chromosome abnormality in women.1 The syndrome is caused by complete or partial absence of the second X chromosome, with or without cell line mosaicism.1

It has been established that most women with Turner’s syndrome do not carry the ‘typical’ karyotype of 45,X, but several different variants of the karyotype, all exhibiting the same clinical features.2 It has been estimated that only about 1% of 45,X fetuses survive to term and that as many as 10% of spontaneous miscarriages have a 45,X karyotype.1

Short stature and gonadal dysgenesis are two characteristic features of the condition, but it is now recognised that the syndrome has a wide variety of features that are well documented in the literature.1 Many of these characteristics result in increased morbidity and mortality, particularly the cardiovascular and metabolic effects. Some typical features allow the presence of the condition to be suspected prenatally on ultrasound and confirmed by prenatal karyotyping. The condition may be suspected when typical features are noted postnatally, but diagnosis is often delayed into childhood, adolescence and adulthood.2

Despite its association with gonadal dysgenesis, 30% of girls affected with Turner’s syndrome will undergo spontaneous puberty.1,2 Concerns regarding future risk of osteoporosis and the emotional consequences of delayed puberty lead many clinicians to commence treatment with estrogen therapy fairly early in adolescence, and so affected girls may not know if they would have reached puberty spontaneously. Attaining spontaneous puberty does not, however, equate with fertility and, where it occurs at all, progressive ovarian failure almost invariably follows.1 This has lead some researchers to raise the possibility of cryopreservation of ovarian tissue from adolescent girls with Turner’s syndrome in order to treat future infertility.1,3-5

Most girls are now treated with a combination of growth hormone, with or without oxandrelone, and HRT.1 Affected individuals require...
ongoing medical supervision throughout their adult life, as
they are at an increased risk of several common diseases,
and a multidisciplinary approach to treatment, preferably in
a tertiary centre, is encouraged.1

Those individuals who undergo spontaneous puberty
and who menstruate may have the potential to achieve
spontaneous pregnancy, although most women affected
will suffer from primary infertility and would require
infertility treatment by oocyte or embryo donation in order
to conceive. Women with Turner’s syndrome who
menstruate normally need to be advised not to postpone
pregnancy because of the risk of early and progressive
ovarian failure.1 There have been many reports of
spontaneous pregnancy in Turner’s syndrome, more
commonly in the mosaic rather than the non-mosaic forms
of the condition.4–14 The prevalence is estimated at
2–7.6%.1,4 Natural pregnancies have been reported in
amennorhoeic women with Turner’s who were receiving
HRT, and in those with raised gonadotrophin levels.1,5,6,8
In those in whom the degree of infertility has not been
established there may be a need for contraceptive advice.5

The COC would seem a reasonable choice given the
increased risk of osteoporosis in these patients;5 however,
the risk of hypertension in these may patients may be a
relative contraindication to the COC.1 Turner’s syndrome is
a relative contraindication to pregnancy. It is known that
there is a increased risk of miscarriage and of stillbirth in
pregnancies of affected individuals, part of chronosomal
abnormality or congenital malformation in any offspring,
and so genetic counselling may be necessary.1,4,5,7,10,12
Oocyte donation and embryo transfer avoids some of these
risks and is now an accepted treatment for infertility in
Turner’s syndrome.1,4 In addition, women with Turner’s
syndrome have an increased cardiovascular morbidity and
mortality (particularly related to aortic dissection) during
pregnancy, and this may occur in natural pregnancies or
those resulting from oocyte donation.1,4,9,14,15 The risk of
death in women with Turner’s syndrome during pregnancy
associated with assisted conception has been estimated as
100 times the normal risk.15 Counselling on safe sex and
the prevention of STIs, and the need for cervical cytology,
are just as important for those with Turner’s syndrome as
for any other sexually active woman.

In our patient the diagnosis was delayed as the only
obvious feature of her condition was short stature, and it
was not until this was investigated that the diagnosis was
made. EE was commenced at the age of 11 years, so that
there was no opportunity for the patient to attain
spontaneous puberty, and she had never menstruated
spontaneously. However, she has had regular withdrawal
bleeding when taking EE and norethisterone, and when
taking the COC. She had assumed that she would be
infertile and for this reason had never sought contraceptive
advice. Although she was sexually active, she did not
practise safe sex, and therefore was at high risk of
developing STIs. Her elevated LH and FSH levels are in
keeping with ovarian failure although there is the possibility
of ovarian resistance. Spontaneous pregnancy in a patient
with Turner’s syndrome with elevated gonadotrophin levels
has been reported.5 In addition to her sexual health needs,
the patient needs to be under regular review because of
other possible associated health problems such as
hypertension, cardiovascular disease, diabetes mellitus and
osteoporosis. Many patients affected by Turner’s syndrome
tend to drift away from medical supervision as they are
discharged from paediatric clinics in their teenage years.
This appears to have been the case with our patient. The
importance of this transition period in the management of
Turner’s syndrome has been recognised.1,16,17

Conclusions
Young women with Turner’s syndrome should not
routinely be assumed to be infertile. Spontaneous
pregnancy, although rare, can occur and carries risks for the
fetus and the woman concerned. Fertility is an important
issue in the management of women with Turner’s
syndrome and while many individuals will need assisted
conception, there are also some who will require
contraceptive advice. As with any other sexually active
woman, they should be advised regarding safe sex and the
need for cervical smear tests. Women who are considering
a pregnancy require preconceptual advice and genetic
counselling, as well as full medical evaluation.

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