The Patient with Turner’s syndrome: Care in the Adult Clinic

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Talk outline

- The care of the adult patient with Turner’s syndrome
- Medical problems which may be encountered
- Transition care
- Be aware of health needs
Disclaimer!

Every woman with Turner’s Syndrome is unique.

No generalisation will apply to everyone.
Turner’s syndrome in the adult clinic

- Patients with known Turner’s syndrome
- New diagnosis
- Manage existing health problems and screen for new ones
Age at diagnosis of patients with Turner’s Syndrome

Median age at diagnosis:
45,X: 13.3 (95% CI: 12.1 - 14.2) years
Iso Xq: 14.2 (12.4 - 16.2) years
Other karyotypes: 19.1 (17.8 - 21.9) years

Stochholm K et al. JCEM 2006;91:3897-3902
# The adult with Turner’s syndrome: many body systems may be affected

<table>
<thead>
<tr>
<th>Body System</th>
<th>Condition</th>
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<tbody>
<tr>
<td>Heart</td>
<td>Ovarian function</td>
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<tr>
<td>Kidneys</td>
<td>Psychosocial</td>
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<tr>
<td>Hearing</td>
<td>Thyroid</td>
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<tr>
<td>Skin</td>
<td>Diabetes</td>
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<tr>
<td>Bones</td>
<td>Liver</td>
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<tr>
<td>High blood pressure</td>
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</tbody>
</table>
Multidisciplinary team

- Endocrinologist
- Gynaecologist
- Cardiologist
- Audiologist
- Psychologist
- Other specialists
<table>
<thead>
<tr>
<th>Feature</th>
<th>Frequency (%)</th>
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<tbody>
<tr>
<td>Short stature</td>
<td>98</td>
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<tr>
<td>Gonadal failure</td>
<td>95</td>
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<tr>
<td>Osteoporosis</td>
<td>50</td>
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<tr>
<td>Cardiovascular defects</td>
<td>25-40</td>
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<tr>
<td>Carbohydrate intolerance</td>
<td>30-40</td>
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<tr>
<td>Hypertension</td>
<td>25-40</td>
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<tr>
<td>Structural kidney abnormalities</td>
<td>35</td>
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<tr>
<td>Hypothyroidism</td>
<td>35</td>
</tr>
<tr>
<td>Multiple naevi</td>
<td>25</td>
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</tbody>
</table>
Monosomy
45XO
V
Mosaic
THE HEART
The heart

• Congenital heart disease
  – Bicuspid aortic valve
  – Aortic co-arctation
  – Aortic root dilatation
• Ischaemic heart disease
• High blood pressure (Hypertension)
The Heart

Aortic co-arctation (narrowing)

Bicuspid aortic valve
Aortic root dilatation
AORTIC DISSECTION

Diagram showing aortic dissection with labels:
- Aorta
- Blood in wall of artery
- Thoracic aorta
- Abdominal aorta

Comparison between normal and dissection states.
Detecting heart abnormalities

- Blood pressure
- Heart exam
- ECG
- ECHO
- MRI heart
High blood pressure

- Anti-hypertensives
- Aim 130/80 mmHg
Risk factors for ischaemic heart disease

- High blood pressure
- Diabetes Mellitus
- Lipid abnormalities
- Oestrogen deficiency
Recommendations

- Yearly review of blood pressure
- Echo and MRI heart 5 - 10 yearly
  - Before pregnancy
  - Onset of hypertension
  - Referral to cardiologist if abnormal

Severe chest pain, even if it doesn’t last long, go to an Emergency Department immediately
Diabetes Mellitus
Diabetes Mellitus

• 4-fold increase in diabetes mellitus

• Up to 50% are insulin resistant

• Obesity common problem

• Abnormal lipid panel – high LDL and hypertriglyceridaemia
Diagnosis of Diabetes

- Fasting glucose
- Oral glucose tolerance test
- HbA1c
Management of diabetes

- Healthy eating
- Exercise
- Oral hypoglycaemic medications
- Treat high blood pressure
- Treat abnormal lipids
The Thyroid
Thyroid Disease

- Increases with age
- 25 to 30 percent hypothyroidism
- Yearly evaluation of thyroid function is recommended
Underactive thyroid (hypothyroidism)

- Symptoms: constipation, low energy, dry skin and weight gain
- Thyroid blood tests
- Thyroid Antibodies
- Treatment: Eltroxin
Overactive thyroid (hyperthyroidism)

- Symptoms: weight loss, heart racing, loose bowel motions, heat intolerance

- Thyroid blood tests
- Thyroid antibodies

- Treatment: Medication/Radioactive iodine/Surgery
Ovarian Function
Ovarian failure

• Ovaries not capable of producing female hormones and eggs.

• Delayed or absent pubertal development

• Primary Amenorrhoea

• If spontaneous puberty, risk of premature menopause
Hormone replacement therapy

• Needed for healthy bones, development of secondary sexual characteristics.

• Prescribe until natural age of menopause.

• Oral or transdermal.
Reproduction

- Few women will maintain sufficient ovarian function to conceive.
- Spontaneous pregnancy occurs in less than 5%
- In live born, risk of Chromosomal abnormalities.
Fertility options

- Adoption.
- In-vitro fertilisation with egg donation by a related or anonymous donor or donor embryo transfer.

- Essential for thorough physical evaluation (particularly cardiac and renal) before attempting pregnancy.
Bone health
Osteoporosis

• Two to threefold increase in the risk of fracture.
• Baseline DXA.
• Technical difficulties in measuring Bone mineral density (BMD)
• If BMD is normal, additional follow-up is done when the patient stops oestrogen.
Management

- Stop smoking
- Reduce Alcohol
- Weight bearing exercise
- Adequate Vitamin D/calcium
- Hormone replacement therapy
Liver and bowel
Gastro-Intestinal system

- Abnormal Liver Function Tests
- Inflammatory bowel disease
- Coeliac Disease: 4 to 6 percent of patients affected
- Intestinal telangiectasia
Coeliac disease

• Reaction to gluten, a protein in the diet
• Gluten is found in wheat and related grains, including rye, barley and possibly oats.
• Symptoms
  – Recurring abdominal bloating and pain
  – Chronic diarrhea or chronic constipation
  – Weight loss not consistent with the amount of food consumed
  – Pale, foul-smelling stool
  – Unexplained anaemia (low count of red blood cells)
• Diagnosis: initial blood test and then biopsy of small intestine and OGD
• Treatment: gluten free diet
Kidney

Anatomy of the Kidney

- Calyces
- Renal Pelvis
- Renal Artery
- Medulla
- Renal Vein
- Ureter
- Cortex
Kidney

- Congenital renal abnormalities 9-times more common – horseshoe kidney
- Renal vascular abnormalities
Skin
Naevi

• ‘Moles’
• Raised and palpable
Hearing and vision
Hearing

• Early sensorineural (nerve) hearing loss

• Check hearing regularly throughout life.

• Protect hearing by avoiding loud noises and using ear protection in loud environments.
Psychosocial aspects

• Adaptation problems
• Memory, attention deficits
• Visual processing
• Maths and geometry – reading maps and following directions
• Concentration difficulties
Transition care

- The transitional period from paediatrics to adulthood
- Consistent medical care
- Screening – detecting previously unrecognised medical problems

- Follow up in adulthood essential
Clinic for patients with Turner’s syndrome

**Medical diagnoses:**
Karyotype:

**Medications:**

**Assessment:**

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<thead>
<tr>
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<tr>
<td>Echo: date and result</td>
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<td>MRI Heart:</td>
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<td>TFT s and Thyroid autoantibodies:</td>
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<td>Fasting glucose, HbA1c, lipid profile, renal, liver, bone profiles:</td>
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<tr>
<td>TTG:</td>
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<td>DXA:</td>
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<td>Audiogram:</td>
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<td>Renal and Pelvic US:</td>
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**Annual follow up**

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**Three to five yearly**

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<td>Fasting glucose, lipid profile, renal, liver, bone profiles</td>
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<td>Bone densitometry (if abnormal)</td>
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Written information on Turner’s syndrome
Summary

• Distinct group of patients
• Specific medical needs
• Endocrinologist as co-ordinator – Multidisciplinary team approach
• Individualise treatment
• More research required
Thank you!