CARDIAC ASSOCIATIONS WITH TURNER SYNDROME

Dr Carla Canniffe,
Specialist Registrar in Cardiology.
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Background

Cholesterol & Blood Pressure

Bicuspid Aortic Valve

Aortic Coarctation

Aortic Dilatation

Aortic Dissection
Background

- Congenital heart disease
  - Range 14-77%
  - Large range of conditions

- Acquired heart disease
  - HTN
  - Cholesterol
  - Ischaemic heart disease
<table>
<thead>
<tr>
<th>Karyotype</th>
<th>This Study*</th>
<th>Gotzsche et al(^3)</th>
<th>Mazzanti et al(^4)</th>
<th>Hou et al(^5)</th>
<th>Parchment et al(^6)†</th>
<th>Moore et al(^7)</th>
<th>Gianzo et al(^8)</th>
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<tbody>
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<td>45,X</td>
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<td>2/4 (50)</td>
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<td>11/62 (18)</td>
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<td>Total</td>
<td>96/244 (39)</td>
<td>46/179 (26)</td>
<td>9/55 (17)</td>
<td>11/49 (22)</td>
<td>7/23 (30)</td>
<td>9/20 (45)</td>
<td>5/19 (26)</td>
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* Does not include conduction defects, MVP, pericardial effusion, pulmonary artery kinking.
† A total of 63 individuals were evaluated. Karyotypes were only given for those groups in which malformations were found.
‡ For example, 45,X/47,XXX; 45,X/46,XX/47,XXX; 45,X/47,i(Xq), i(Xq); 45,X/46,X + complex rea(X).
Number of affected individuals over the number of individuals evaluated with the same chromosome complement. Percentages are given in parentheses.
Acquired Cardiac Conditions
Cholesterol

Bad vs. Good Cholesterol

Bad (LDL)
stores cholesterol in the blood stream

Good (HDL)
regulates LDL storage and promotes excretion

Atherosclerotic Plaque (LDL accumulation)
Cholesterol

- 37-50% abnormal lipid profile
- Oestrogen
- X-linked gene effect

Compared to women with POF, Turner's had ↓ HDL and ↑ LDL & Triglycerides.

Difference in particle size
Cholesterol

- Management
  - Low fat Diet
  - Regular Exercise
  - Healthy weight
  - Take medication if needed
  - Annual screening
Hypertension
Hypertension

- Systolic hypertension raised compared to age matched peers

The ABPM suggests optimal 24-hour blood pressure (113 mmHg / 76 mmHg daytime, 96 mmHg / 61 mmHg night-time).
Hypertension

- Appears to be more common in 45XO.
- 30% of those with hypertension have underlying cardiac abnormality

Management
- Regular Screening & follow up
- Medication
- 24 hour ABP...right arm if history of aortic coarctation
Ischaemic Heart Disease

- Dyslipidaemia
- Hypertension
- Diabetes
- Obesity
- Oestrogen deficiency
- Abnormal vessel structure
Congenital Cardiac Conditions
Bicuspid Aortic Valve
Bicuspid Aortic Valve

- 1-2% general population v 12-47% in TS population.
- More common in 45XO.
- Limited follow-up data in TS groups.

General population;
  - Aortic stenosis commonest valve problem.
  - 52% develop aortic dilatation.
  - Incidence of surgical intervention 22-27%. 
Bicuspid Aortic Valve

- **ECHO**
  - Valve leak/ tightening?
  - Effecting heart muscle?
  - Dilatation in Aorta
  - +/- MRI

- Exercise advice dependent on function & aorta measurements.
- Medication as required.
- Surgical Intervention maybe required.
Aortic Coarctation
Aortic Coarctation

- 0.2-0.6 per 1,000 live births in general population.
- TS range 10-45% - more common in 45XO
Aortic Coarctation

- Detection & Screening
  - Pulse check.
  - Upper and lower limb BP
  - All TS echo at time of diagnosis and CMR once able to lie flat.

- Repair
  - Surgical
  - Stent

- Follow-Up
  - BP control
  - Annual specialist review + imaging
Aortic Dilatation
Aortic Dilatation

- Associated with other cardiac conditions
  - BAV
  - Coarctation
  - HTN

- Less clear
  - Rate of dilatation
  - Cut-off values – INDEX TO BODY SIZE
Aortic Dilatation

- Management

- ASI > 2 cm/m² significant dilatation requiring close surveillance +/- medication

- ASI ≥ 2.5 cm/m² extreme dilatation

- ASI ≥ 2.5cm/m² + Absolute AD of ≥ 3.5cm used as cut-off for prophylactic intervention
Aortic Dilatation

- Screening & Follow Up
  - ECHO
    - 5 yrs if normal
    - Annual if cardiac abnormality or dilatation
    - 6 monthly if concerns about increasing measurement
  - MRI
    - At least once & at appropriate times

- Compare like with like
Aortic Dilatation

Management

- Aggressive BP control
- B-Blockers/ ARBs
- Frequent Imaging
- Exercise restriction
- Prophylactic surgical repair
Aortic Dissection

[Diagram showing normal and dissected aorta]
Aortic Dissection

<table>
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<tr>
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<th>Turner Syndrome</th>
<th>General Population (Female)</th>
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<tr>
<td>Incidence</td>
<td>36-78 per 100,000 pt yrs</td>
<td>9 per 100,000 pt yrs</td>
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<tr>
<td>Average age (years)</td>
<td>31.5</td>
<td>68</td>
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<tr>
<td>Associated CHD (%)</td>
<td>70-95</td>
<td>20 CTD</td>
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<tr>
<td>HTN</td>
<td>47%</td>
<td>44-75%</td>
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Aortic Dissection

- International Turner Syndrome Aortic Dissection Registry 2005-2010

- Presenting Symptoms:
  - Chest Pain (16/19)
  - Neck/ back Pain (6/19)
  - Abdominal Pain (1/19)
  - SOB (5/19)
  - Nausea/ vomiting (5/19)
  - Dizzyness/weakness (4/19)
  - Impending doom (2/19)
Aortic Dissection

- 10-30% no pre-disposing risk factors identified.
  - BAV
  - Aortic coarctation
  - Aortic dilatation (ASI >2.5cm/m2)
  - Elongated aortic arch
  - 45XO (54% cases)
  - Increasing Age
  - HTN
  - Pregnancy
Aortic Dissection

- Awareness and Education
  - Risk factors
  - Early presentation with symptoms

- Management
  - Aggressive BP control
  - Frequent Imaging
  - Exercise restriction
  - Prophylactic surgical repair
General Advice
Cardiac Screening & Follow Up

- Baseline cardiac evaluation at time diagnosis
  - Blood pressure (all limbs)
  - ECG
  - Echo
  - MRI (older/ sedation if high clinical suspicion)

- Follow up depends on clinical situation
  - Normal CVS and age-appropriate BP
    - Re-evaluation at timely occasions e.g. Transition to adult clinic, before attempting pregnancy or if HTN develops
    - Imaging every 5-10 years.
Follow Up

- Cardiac abnormalities
  - Dependent on abnormality

- 6/12 BP check with GP
- Annual specialist review + imaging
Pregnancy

- Clinical practice guidelines Care of Girls and Women with Turner Syndrome
  - Hypertension, Aortic dilatation, Ao Coarctation +/- BAV relative contraindications.
- American Society of Reproductive Medicine
  - Cardiac anomaly absolute contraindication

- Increased risk of dissection.
- 40% normal fetal-maternal outcome.

- Screening pre-pregnancy & counselling as appropriate
- Close cardiac monitoring throughout pregnancy & post-partum period.
General Advice
If I present with chest pain, please note, aortic dissection may occur in certain patients with Turner Syndrome.

www.tcgi.ie

“I HAVE TURNER SYNDROME”
Please see reverse for medical information

Name: ________________________ DOB: ________________________
Doctor: ________________________ Tel: ________________________
Emergency Contact: ________________________ Tel: ________________________
CARDIAC AND AORTIC DISEASE IN TURNERS SYNDROME

- Mater Misericordiae Hospital

- Looking at cardiac outcomes of Irish patients ≥ 16 years old with TS
  - Cardiac Imaging
  - Presence of HTN +/- medication
  - Aortic Valve or Root surgery
  - Dissections
**CARDIAC AND AORTIC DISEASE IN TURNERS SYNDROME**

Mater Misericordiae Hospital

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<td>DOB:</td>
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I confirm that I am happy to be contacted to get further information about possibly participating in this research project.

Signature:

Date:
Questions?