

CARDIOVASCULAR HEALTH FINDINGS FROM SURVEYS 1- 3

Our current analysis uses data from the Turner Syndrome Research Registry (TSRR) cardiovascular questions asked in Surveys 1- 3. The REDCap export contained **1,623 survey records**, representing **1,042 unique participants**. Because some people completed more than one survey, we collapsed all responses to **one record per participant**.

To focus on individuals who actually provided cardiovascular information, we **excluded 122 participants** who did not answer any of the cardiovascular questions in Surveys 1 or 3. This left an analytic sample of **920 participants** with at least one cardiovascular response.

- The **overall pie chart** in this report is based on these **920 participants**.
- The **stacked horizontal bar chart by karyotype** is restricted to participants who also had a usable karyotype classification. After excluding those with missing or “skipped/NS” karyotype information, the stacked chart includes **629 participants**: 45,X only (n = 272), any mosaic karyotype (n = 117), did not know / no test (n = 190), and other specific karyotypes (n = 50).

Key findings (all karyotypes; n = 920, pie chart)

Distribution of cardiovascular issues — unique participants
Percent of cardiovascular diagnoses

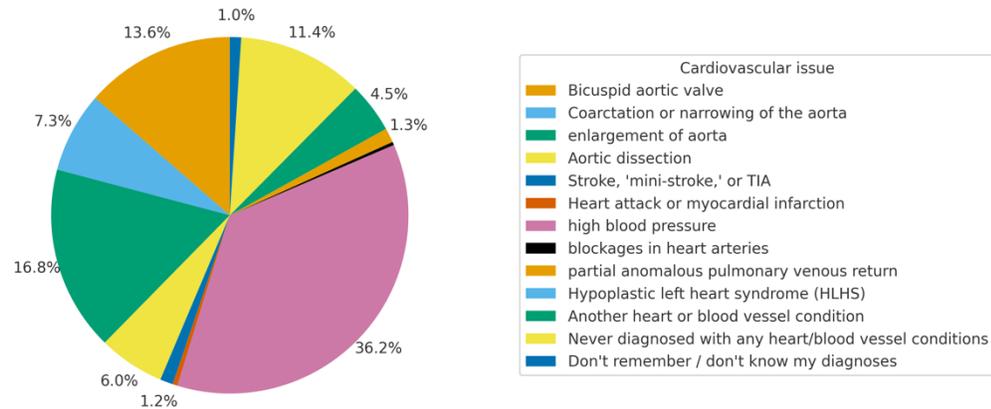


Figure 1: Distribution of cardiovascular diagnoses reported in Surveys 1–3 among 920 unique TSRR participants with cardiovascular data

The overall pie chart shows how these 920 responses are distributed across specific cardiovascular issues:

- **High blood pressure** was the single most common issue, accounting for **36.2%**.
- **Enlargement of the aorta** made up **16.8%**, and the **bicuspid aortic valve** contributed **13.6%**. Together, these aortic/valve problems represented nearly half of all recorded cardiovascular issues.
- **“Never diagnosed with any heart/blood vessel conditions”** accounted for **11.4%**, indicating that a minority of participants with cardiovascular data reported no known diagnoses.
- **Coarctation or narrowing of the aorta** made up **7.3%**, and **“another heart or blood vessel condition”** contributed **4.5%**.
- More serious vascular events were less common but still present in the overall distribution:
 - **Aortic dissection: 6.0%**
 - **Stroke, “mini-stroke,” or TIA: 1.2%**
 - **Heart attack or myocardial infarction: 0.4%**
- Additional issues made up smaller slices of the pie:

- **Partial anomalous pulmonary venous return (PAPVR): 1.3%**
- **Blockages in heart arteries: 0.3%**
- **Hypoplastic left heart syndrome (HLHS): 0.0%** in this dataset.
- **“Don’t remember / don’t know my diagnoses”** accounted for **1.0%**.

These proportions come directly from the **overall pie chart**, which displays the **percent of all recorded cardiovascular responses** attributed to each issue.

Findings by karyotype group (stacked horizontal bar chart; n = 629)

The stacked horizontal bar chart shows the distribution of cardiovascular issues within four karyotype groups: **45,X only (n = 272)**, **any mosaic karyotype (n = 117)**, **did not know / no karyotype testing (n = 190)**, and **other specific karyotype (n = 50)**. Only participants with both cardiovascular data and a usable karyotype classification (**629 participants**) are included in this figure.

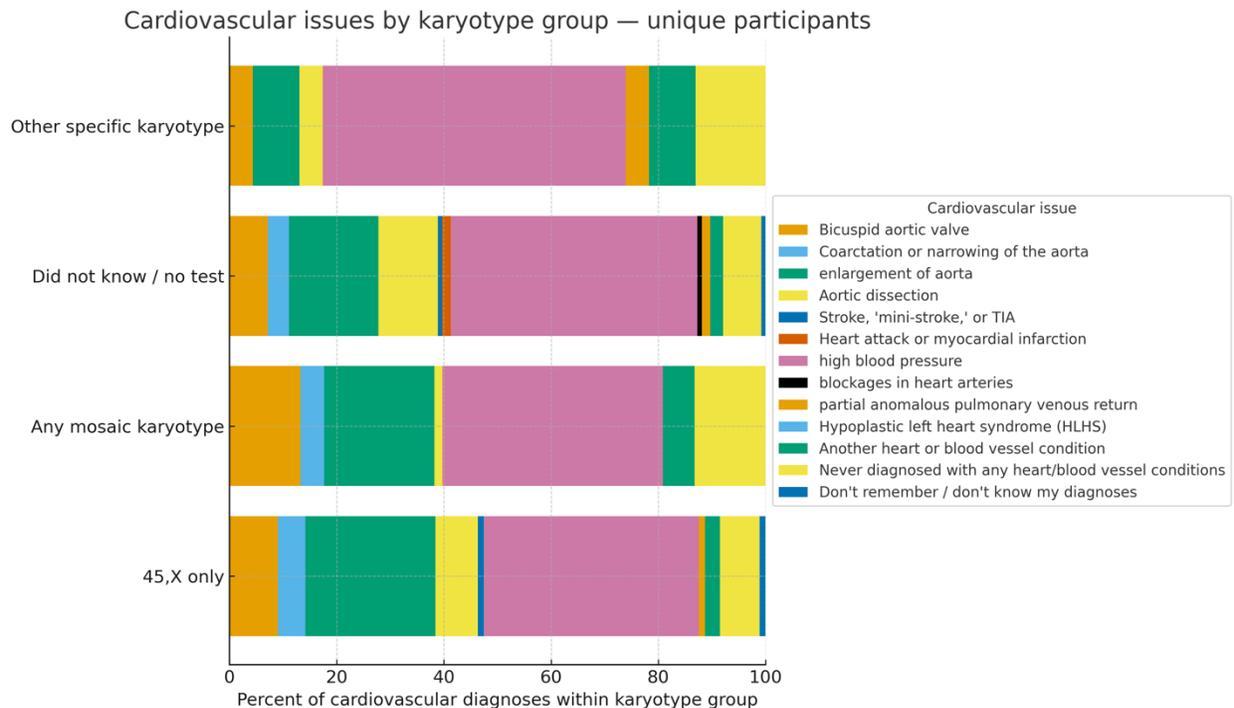


Figure 2: Distribution of cardiovascular diagnoses by karyotype group among 629 TSRR participants with both cardiovascular data and a usable karyotype; percentages sum to 100% within each karyotype group.

Selected patterns from the stacked chart:

- **High blood pressure** was common in every group:
 - 45,X only: **71 of 272 (26.1%)**
 - Any mosaic karyotype: **28 of 117 (23.9%)**
 - Did not know / no test: **58 of 190 (30.5%)**
 - Other specific karyotypes: **13 of 50 (26.0%)**
- **Enlargement of the aorta** occurred across karyotypes:
 - 45,X only: **43 of 272 (15.8%)**
 - Any mosaic karyotype: **14 of 117 (12.0%)**

- Did not know / no test: **21 of 190 (11.1%)**
- Other specific karyotypes: **2 of 50 (4.0%)**
- **Bicuspid aortic valve** and **coarctation of the aorta** were present in all or most groups:
 - Bicuspid aortic valve ranged from **1 of 50 (2.0%)** in other specific karyotypes to **9 of 117 (7.7%)** in the mosaic group.
 - Coarctation of the aorta ranged from **0 of 50 (0%)** in other specific karyotypes to **9 of 272 (3.3%)** in the 45,X only group.
- **Aortic dissection**, while relatively rare, appeared in several groups:
 - 45,X only: **14 of 272 (5.1%)**
 - Any mosaic karyotype: **1 of 117 (0.9%)**
 - Did not know / no test: **14 of 190 (7.4%)**
 - Other specific karyotypes: **1 of 50 (2.0%)**
- Participants who selected “**Never diagnosed with any heart/blood vessel conditions**” made up a small portion of each karyotype group, ranging from **4.8%** in the 45,X only group to **7.7%** in the mosaic group.

Summary

In this analysis, we used data from Surveys 1- 3 of the Turner Syndrome Research Registry to describe cardiovascular health in a carefully defined group of participants. Starting from 1,623 survey records, we identified 1,042 unique individuals and then focused on the 920 participants who answered at least one cardiovascular question. High blood pressure and structural aortic problems (bicuspid aortic valve, enlargement, and coarctation) accounted for a large share of the reported issues, while only a minority of participants indicated that they had never been diagnosed with any heart or blood vessel condition. When we examined these findings by karyotype group among 629 participants with usable karyotype data, we observed cardiovascular concerns across all groups, including 45,X only, mosaic karyotypes, those without karyotype test information, and other specific karyotypes. Overall, these results confirm that cardiovascular surveillance remains a critical component of Turner syndrome care for all TSRR participants, and they highlight ongoing opportunities for blood pressure control, cardiac imaging, education, and follow-up to reduce long-term cardiovascular risk.