

# Isolated Wide CSP: No Increased Risk for 22q11.2 Microdeletion in the Absence of Additional Sonographic Indicators

## Is a wide fetal cavum septi pellucidi associated with DiGeorge syndrome?

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### Objective

- To assess the association between CSP width, length-to-width ratio and 22q11.2 microdeletion

### Methods

- Cross-sectional study
- CSP width and ratio reference Intervals were derived from 138 healthy fetuses with uncomplicated neonatal outcomes (16+0 - 37+0 weeks gestation)
- Nomogram were constructed by using the LMS method
- CSP ratio was calculated by dividing the CSP length by its width.
- Wide CSP : measurement > 95<sup>th</sup> percentile
- 29 fetuses with 22q11.2 microdeletion and adequate CSP visualization from 2014 to 2022 were included
- CSP width measurements and length-to-width ratio were compared to those of the low-risk control population.

### Results

- 75.9% (22/29) such fetuses presented with congenital heart anomalies.
- 13.9% (4/29) of fetuses with 22q11.2 microdeletion had a CSP width above the 95<sup>th</sup> percentile
- No significant difference in CSP z-scores between 22q11.2 fetuses and low-risk group (width + ratio)
- All 4 fetuses with wide CSP had additional anomalies (3 major cardiac anomalies, 1 absent thymus and growth restriction)
- Only 1 of the 4 fetuses had a CSP ratio above the 95<sup>th</sup> percentile
- The remaining 3 fetuses had a normal ratio

### Conclusion

- A wide (>95<sup>th</sup> percentile) but otherwise normal CSP does not indicate 22q11.2 microdeletion

### Figures

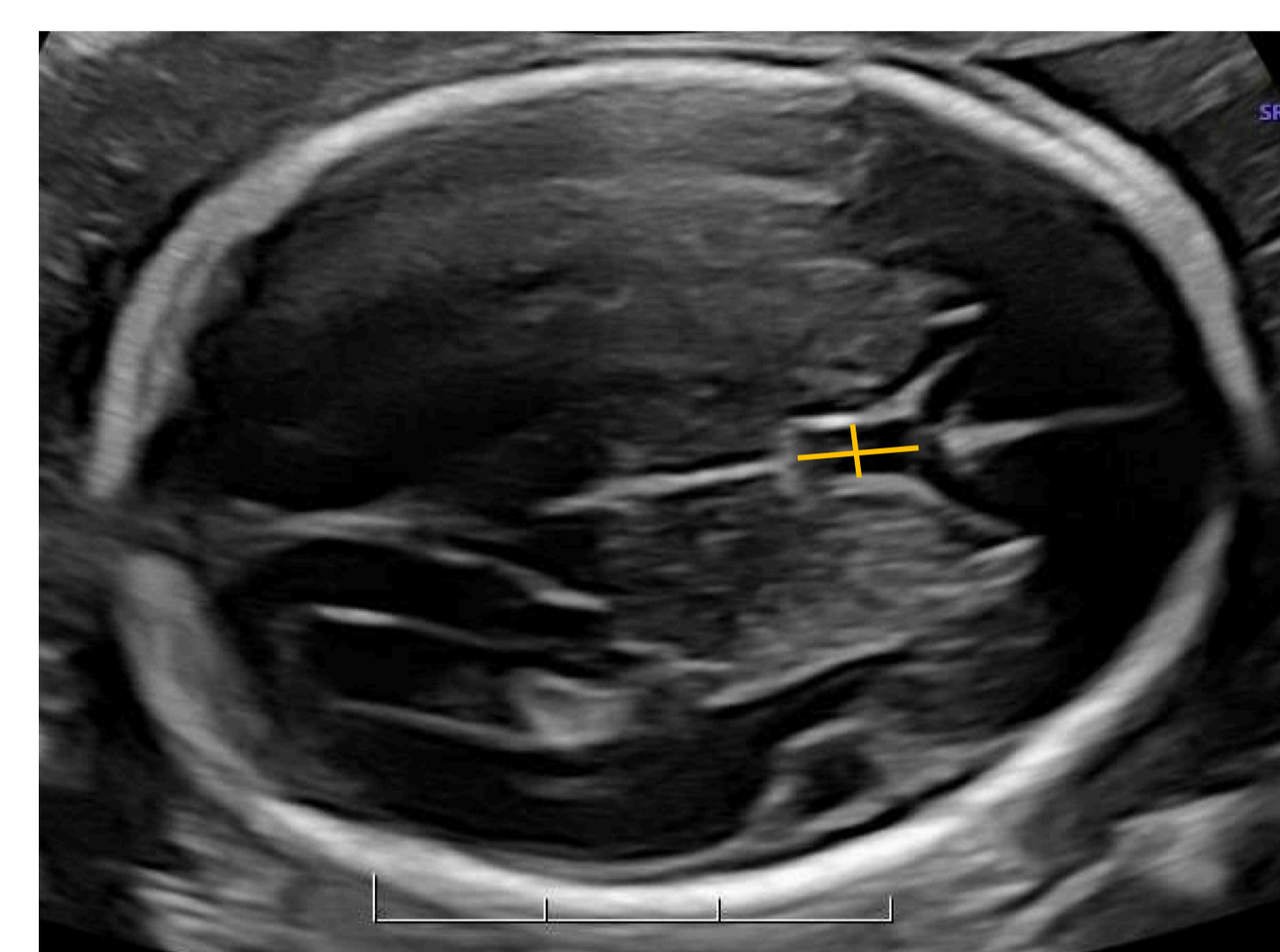


Figure 1. Trans-ventricular axial plane demonstrating the measurement of the CSP width and ratio in a 21+4 week fetus.

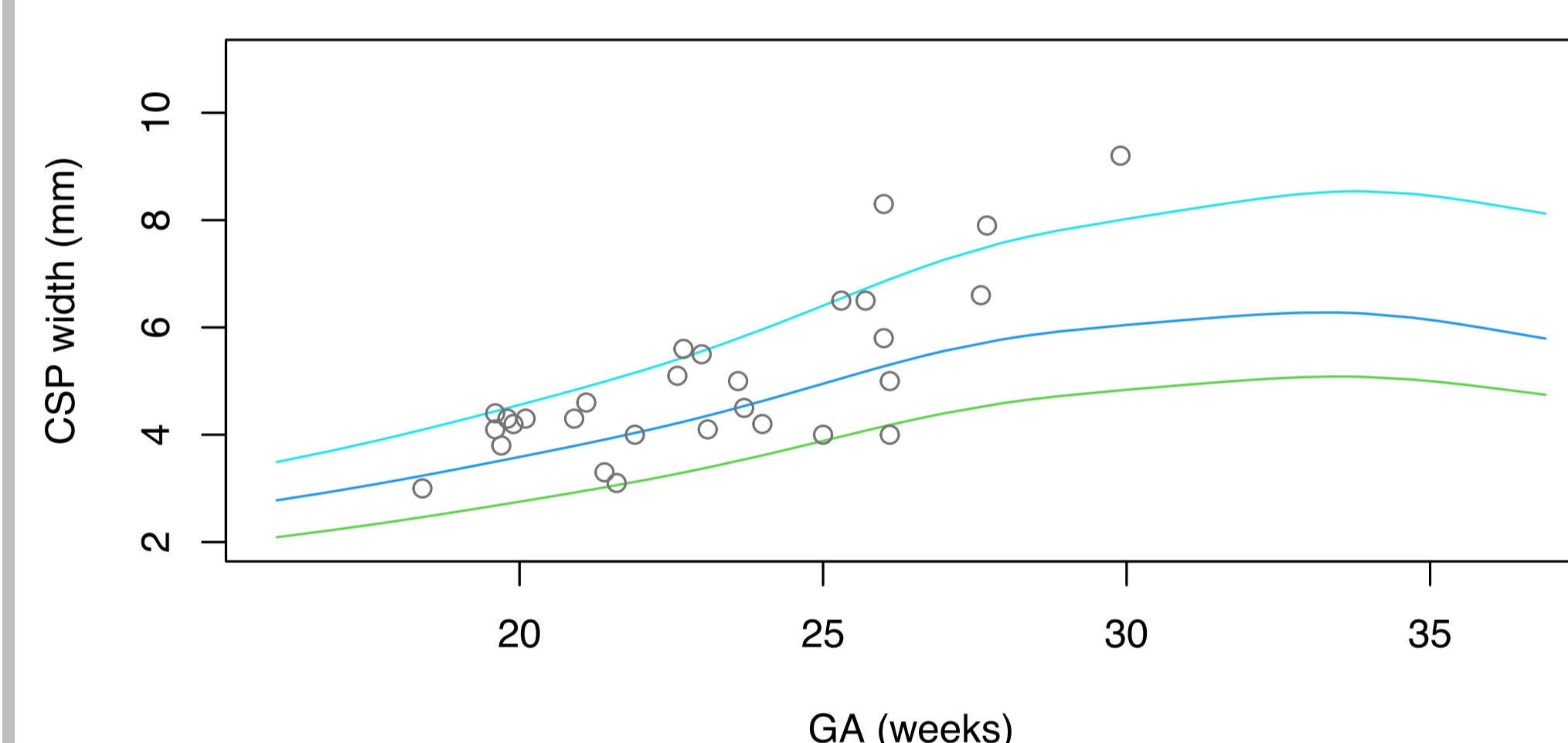


Figure 2. Percentile curves for CSP width, based on LMS method. Lines represent the 5<sup>th</sup>, 50<sup>th</sup> and 95<sup>th</sup> percentiles of the low-risk population. Hollow circles represent the CSP width of fetuses with 22q11.2 microdeletion.

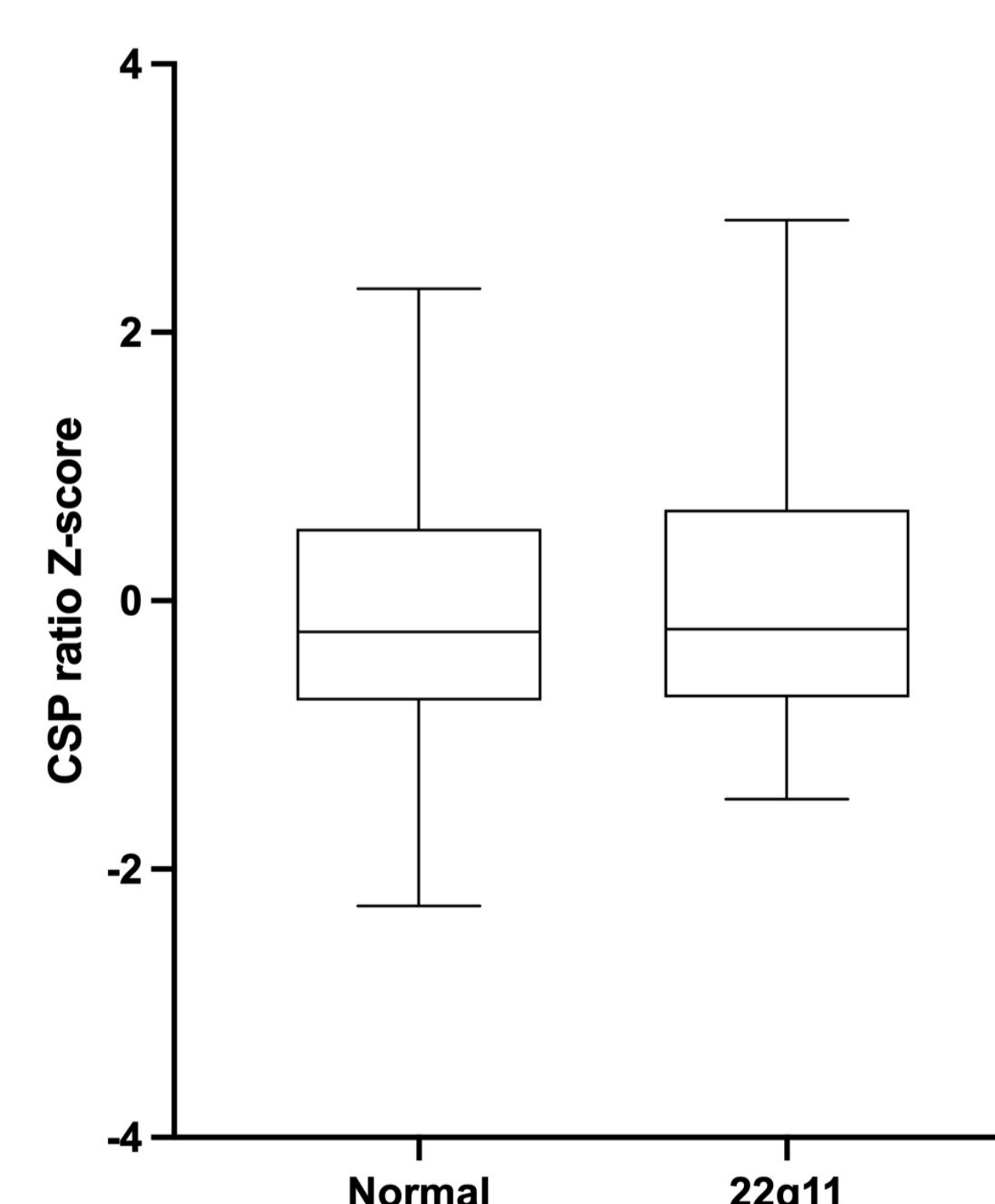


Figure 3. Box-and-whisker plot of the CSP ratio expressed as Z-score in the low-risk and 22q11.2 microdeletion groups. Boxes and internal lines show median and interquartile range and whiskers represent range. No significant difference was found between groups (p = 0.88)