

Monozygosity modifies the anatomical distribution of congenital malformations in twin pregnancies independently of chorionicity

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Objective of the study

- To evaluate the incidence, anatomical distribution, severity, and concordance of congenital malformations in twin pregnancies in accordance with both zygosity and chorionicity.
- To explore if specific anatomical patterns could reflect intrinsic developmental mechanisms related to monozygotic twinning.

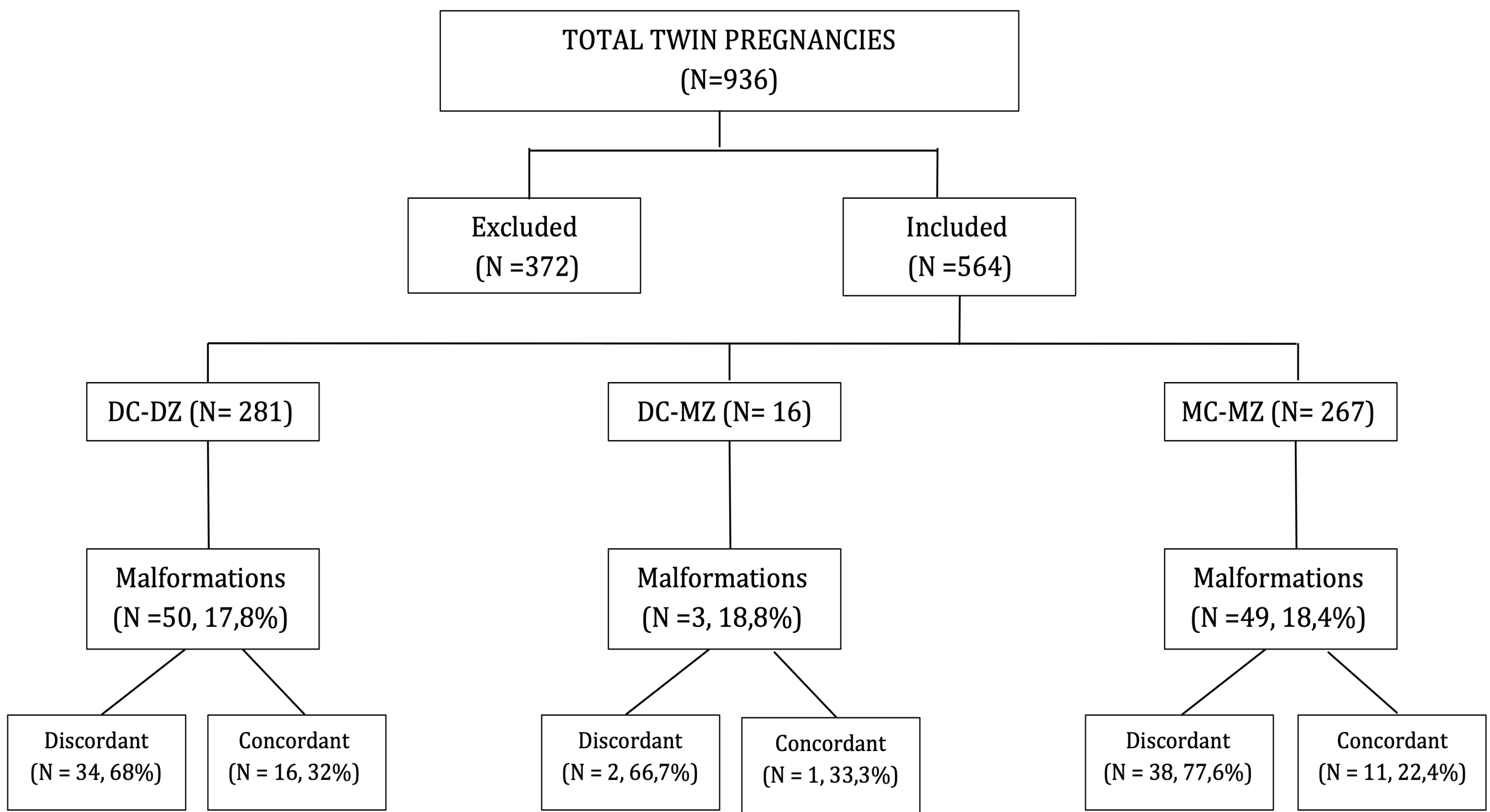
Methods

Retrospective 2016–2025
Single tertiary fetal medicine center
936 twin pregnancies

- ✓ Inclusion criteria:
- Reliable prenatal assessment whit outcome data
 - Chorionicity determined at the 1st trimester
 - Zygosity determination available

- ✗ Exclusion criteria:
- ART
 - Maternal metabolic diseases
 - Secondary hemodynamic MC anomalies

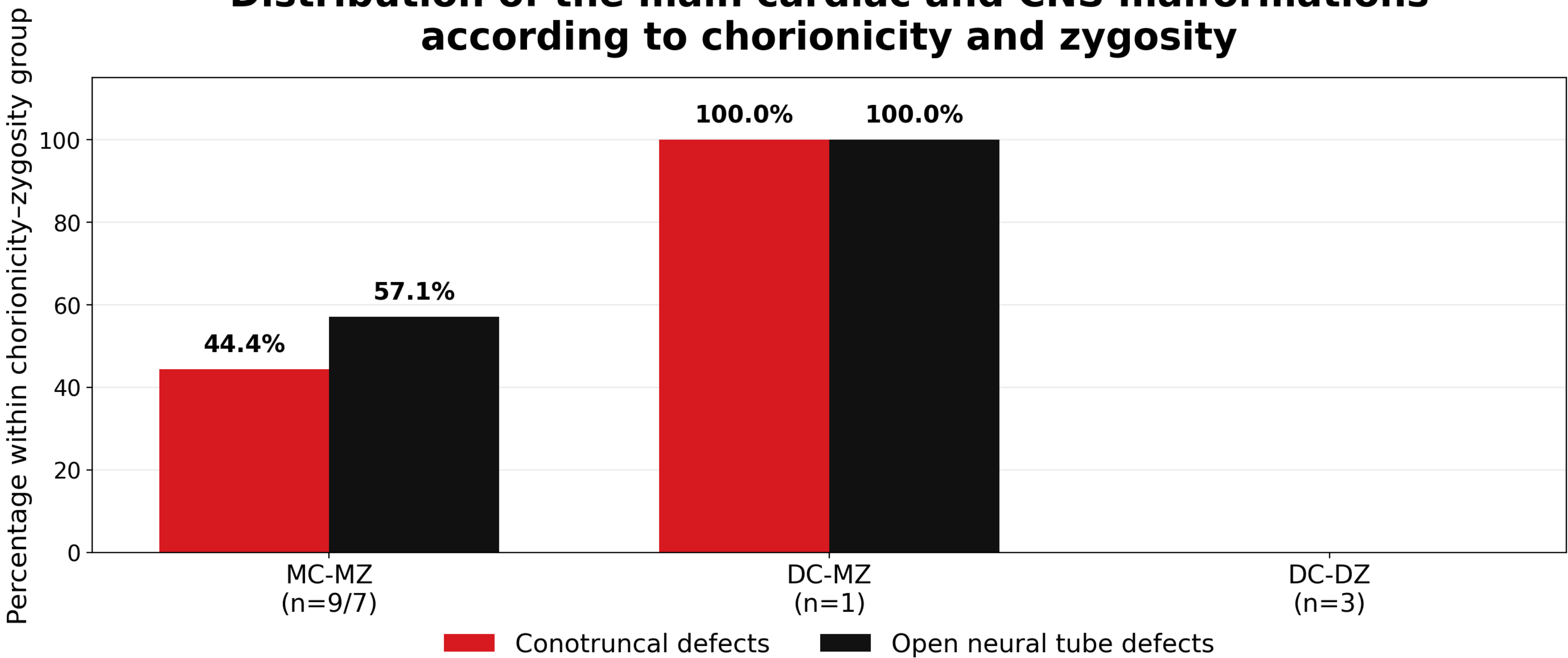
Results



PREGNANCIES WITH CONGENITAL MALFORMATIONS 102
57 MAJOR
45 MINOR

Similar prevalence between groups
Predominantly discordant phenotypes

Distribution of the main cardiac and CNS malformations according to chorionicity and zygosity



MC-MZ: monochorionic monozygotic; DC-MZ: dichorionic monozygotic; DC-DZ: dichorionic dizygotic.

Conclusion

- Monozygosity does not increase the overall prevalence, but selects specific patterns of abnormalities.
- Monozygosity influences the anatomical distribution of malformations depending on early embrionic patterning.
- Conotruncal defects and neural tube defects emerged as the dominant anomalies in monozygotic twins.
- Zygosity should be considered alongside chorionicity when assessing developmental risk in twin pregnancies.

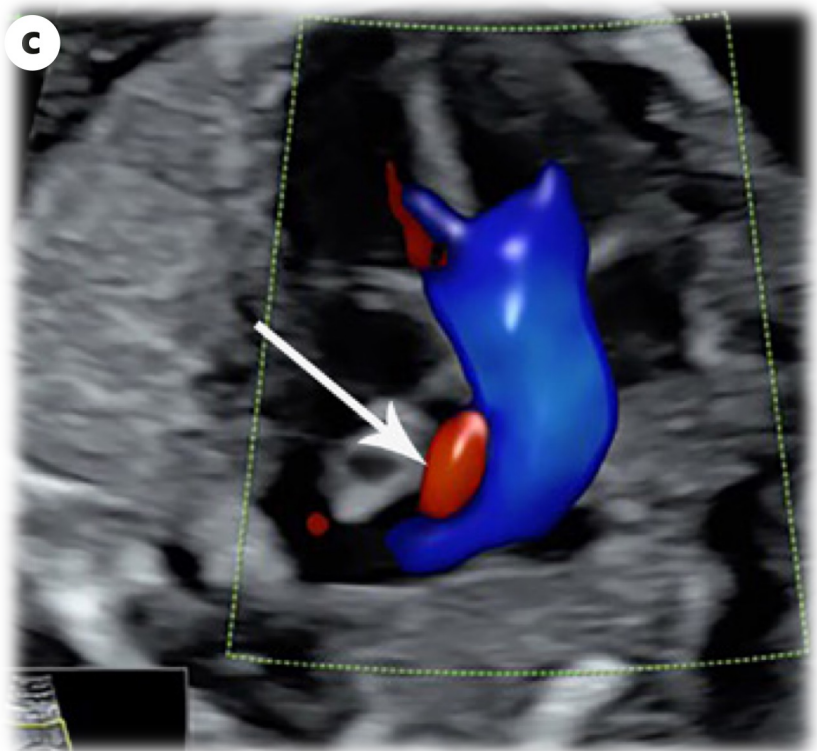
A Developmental Patterning Hypothesis

The development of the cardiac outflow tract and neural tube closure depends on early and coordinated mechanisms of embryonic development particularly vulnerable to initial perturbations.

Monozygotic twinning can represent a transient perturbation of embryonic development at the time of twinning phenomenon or immediately after.

Selective vulnerability of specific developmental fields

Conotruncal defects



+

Neural tube defects

