

EP15.10. Introduction of a nationwide first-trimester ultrasound cohort integrating imaging, clinical, and genetic data for early fetal phenotyping in China

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Purpose

To establish a nationwide, standardized 11–13+6-week fetal ultrasound cohort in China that integrates imaging, clinical, and genetic data, aiming to define early abnormalities, explore genotype–phenotype associations, and enable early risk prediction.

Methods

The Early Pregnancy Fetal Ultrasound Screening Abnormalities Cohort (EPFUS-AC) is a multi-center observational study combining prospective enrollment and retrospective data integration across nine tertiary centers in China.

Standardized ultrasound examinations were performed at 11–13+6 weeks of gestation following unified scanning protocols.

Phenotype recording

During each examination, fetal findings were recorded and classified as soft marker abnormalities or structural anomalies.

Soft markers included NT thickening, cystic hygroma, absent nasal bone, tricuspid regurgitation, and abnormal ductus venosus flow, among others.

Structural anomalies included anencephaly,

Results

As of the latest data update, 168,140 pregnancies were included, comprising both prospective and retrospective data. The cohort captures a broad spectrum of early fetal ultrasound findings, including both soft marker abnormalities and structural anomalies.

Severe first-trimester structural abnormalities were identified, including major cardiac malformations such as single ventricle and lethal anomalies such as anencephaly. Standardized first-trimester ultrasound can therefore detect clinically significant fetal abnormalities at an early stage.

Conclusion

EPFUS-AC is a large-scale, multi-center first-trimester ultrasound cohort with integrated imaging, clinical, and genetic data.

It provides a platform for earlier detection of fetal anomalies and for studying how genetic variation relates to early fetal phenotypes.

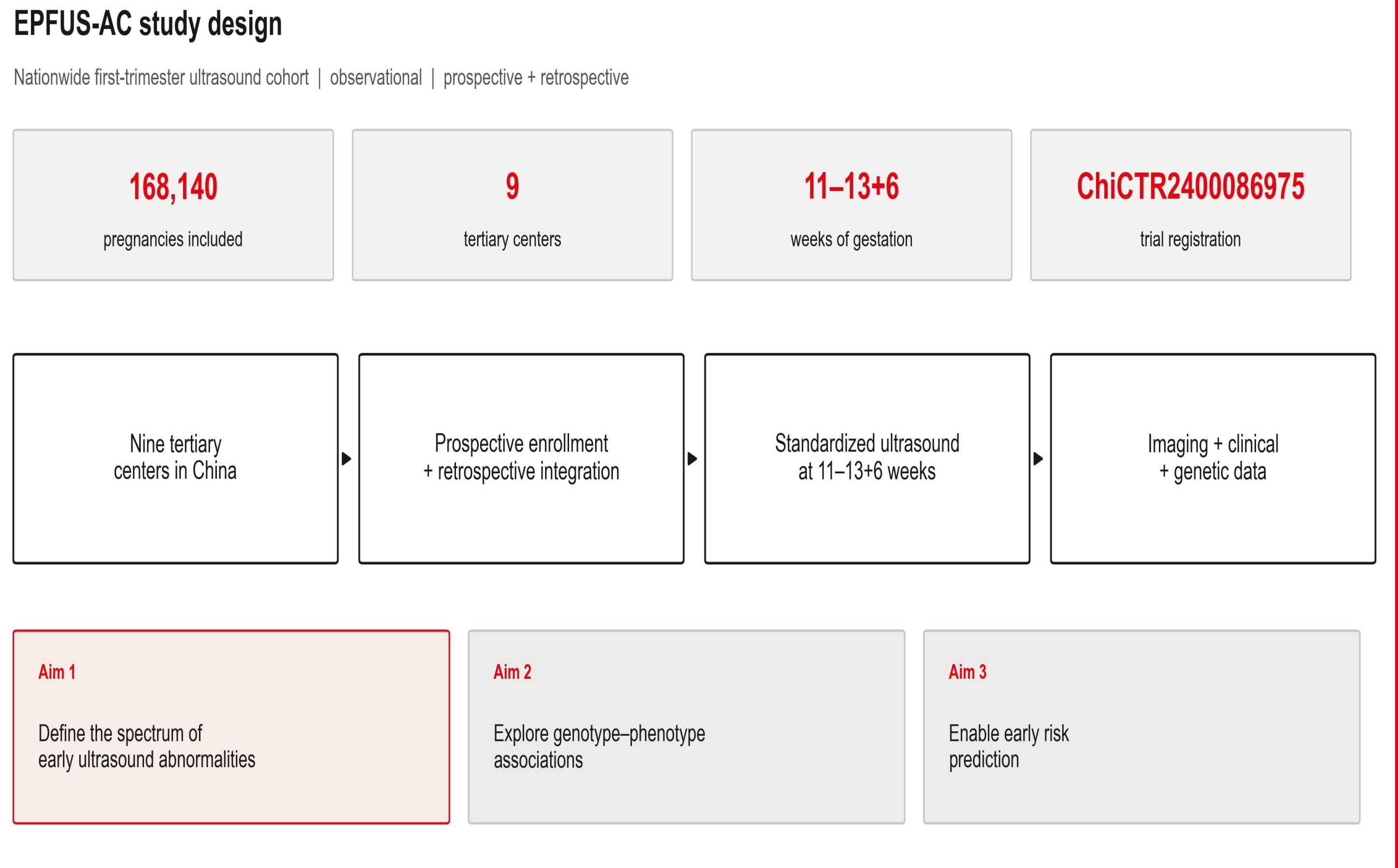


Figure 1. Study design of EPFUS-AC, linking nine-center enrollment, standardized 11–13+6-week ultrasound, and multi-dimensional data (n = 168,140).

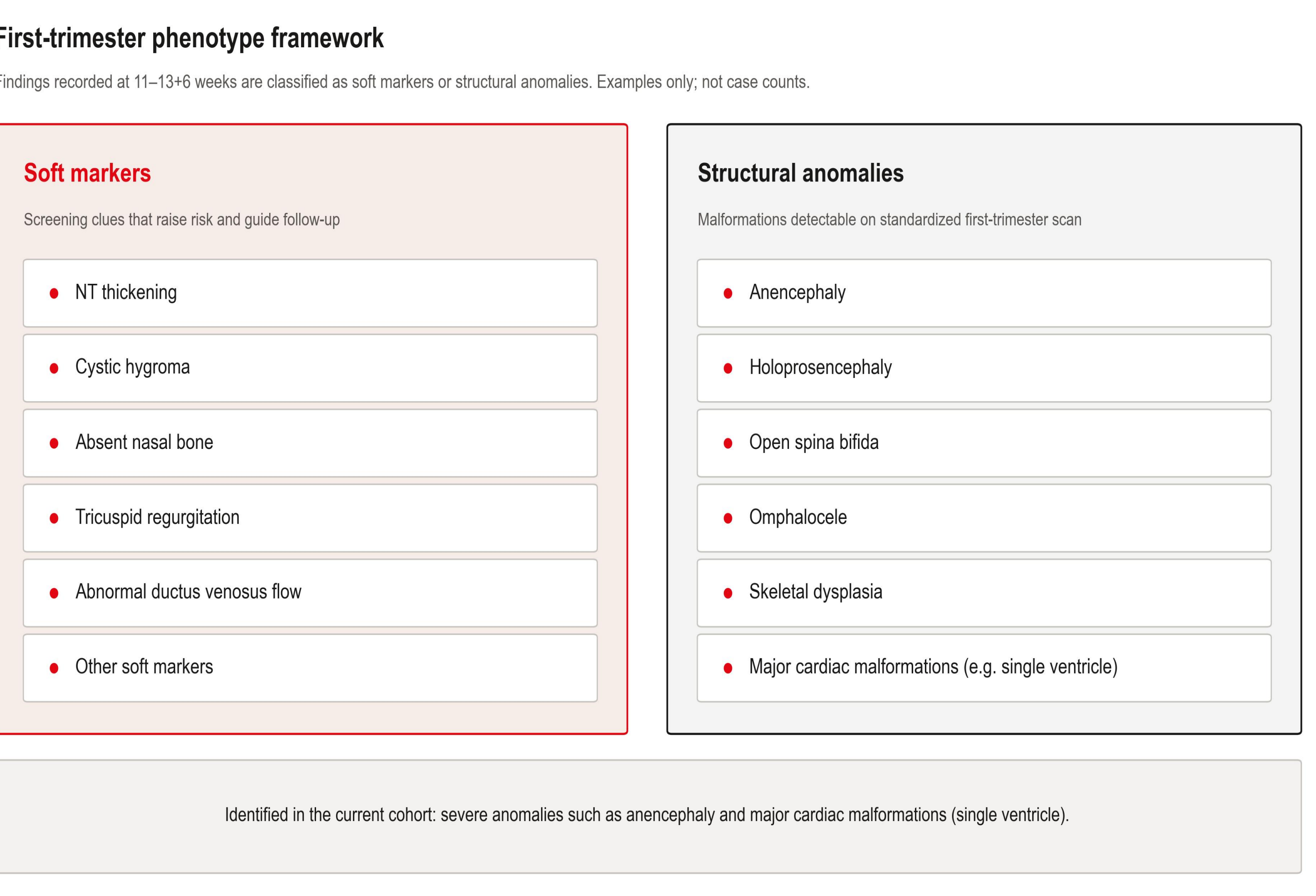


Figure 2. Early phenotype framework used in EPFUS-AC. Listed findings are protocol examples, not frequency counts.