ID: 4469- USE OF EXOME SEQUENCING IN FETUSES WITH COMPLEX CENTRAL NERVOUS SYSTEM ANOMALIES

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Objective:

To describe exome yield of exome sequencing (ES) in a group of fetuses with complex central nervous system (CNS) anomalies.

Methods:

- Retrospective study
- 55 cases of complex CNS
- Between 2017-2021
- Normal array-CGH → Exome sequencing (ES)

Conclusion: ES analysis in complex CNS anomalies reveals abnormalities in about half of the cases, especially in those with associated malformations.

Results:

ES revealed a likely/pathogenic variant in **41.8%** (n=23): 14 AD, 8 AR and 1 X-linked