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Prenatal diagnosis of central nervous system defects and their genetic studies in pregnant women referred to a perinatal reference center in Chile

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Objectives

To describe our experience at a perinatal referral center, about central nervous system malformations (CNS) and the results of invasive prenatal genetic studies in pregnant women from 2017 to 2024.

Methods

Retrospective descriptive study. Pregnant women with genetic study and neurosonography who met the inclusion and exclusion criteria were included.

Results

A total of 172 pregnant women with suspected CNS alteration were recruited. An invasive genetic study was performed in 54% (N= 93), on average at 23 (14-35) weeks of gestational age. Of the total invasive studies, 50% (N=46) were euploid, 41% (N=38) aneuploid, other chromosomal alterations 4% (N=4) and in 5% (N=5) it was not possible to obtain a result.

In 20% (N=10) of the euploid cases, chromosomal microarray analysis or exome sequencing analysis was performed, obtaining 14% (N=7) of point mutations and 6% (N=3) of microdeletions. 75% of the live newborns had postnatal neurological evaluation and 81% of the prenatal brain findings were confirmed by physical examination or postnatal imaging . Some 22% (N=6) of the euploid cases had findings suggestive of genopathy.

Conclusion

In Chile, there are no published studies on genetic testing in pregnant women with malformations CNS. This information allows us to understand the importance of genetic testing in the management and follow-up of these patients. It is also useful to demonstrate the need for more tools for genetic testing in our local reality.

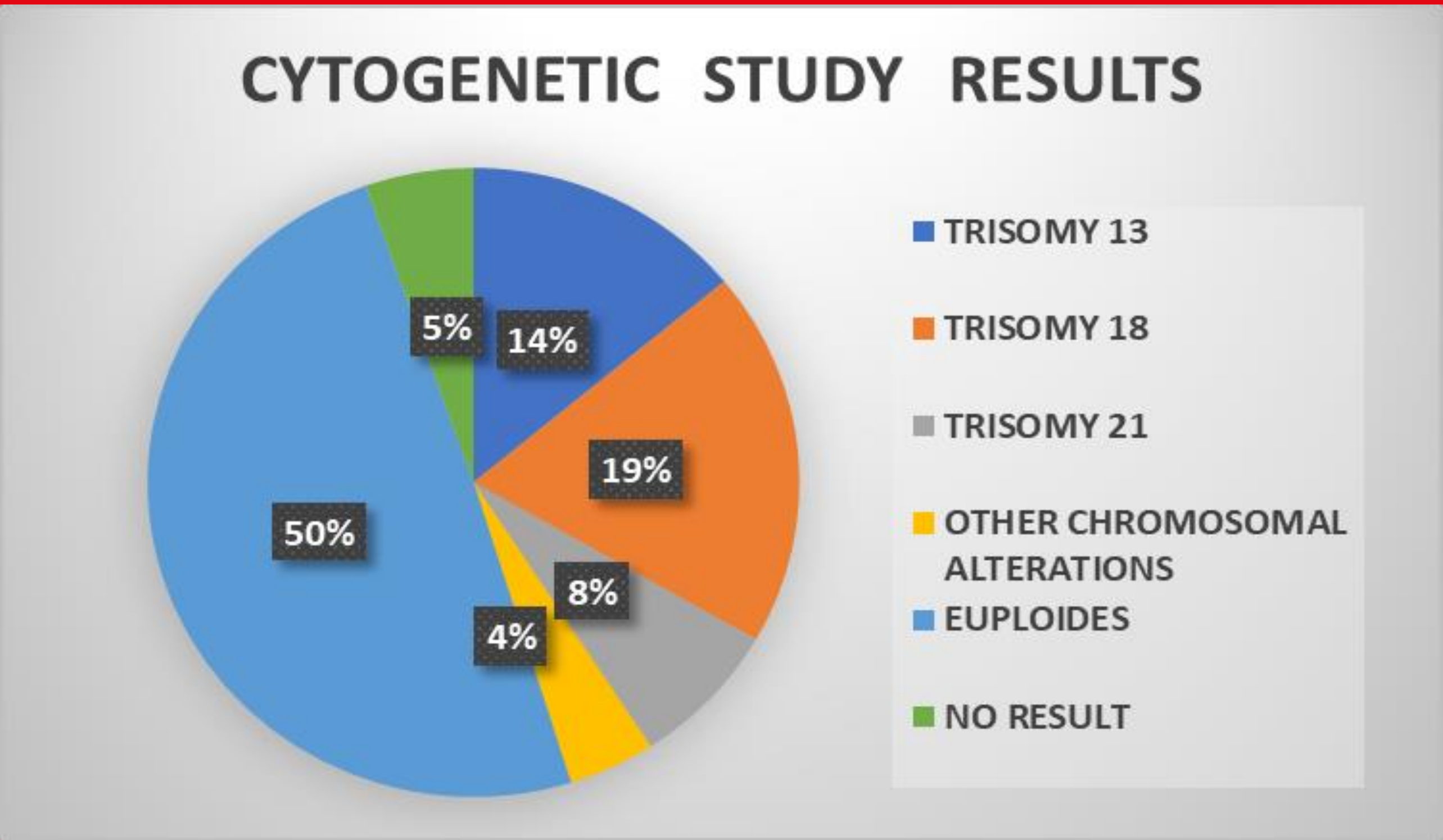


Figure 1. Distribution by cytogenetics study

Genetic Diagnosis	Central Nervous System Malformations (CNS)
Cri Du Chat Syndrome	Posterior fossa malformations
Joubert Syndrome	Dorsal Induction Defects
Mowat Wilson Syndrome	Midline Anomalies
Pathogenic Variant TSC2	Tumor Lesiones
Microdeletion 22q1.1	Midline Anomalies

Tabla 1. Genetic studies and correlation with malformations CNS

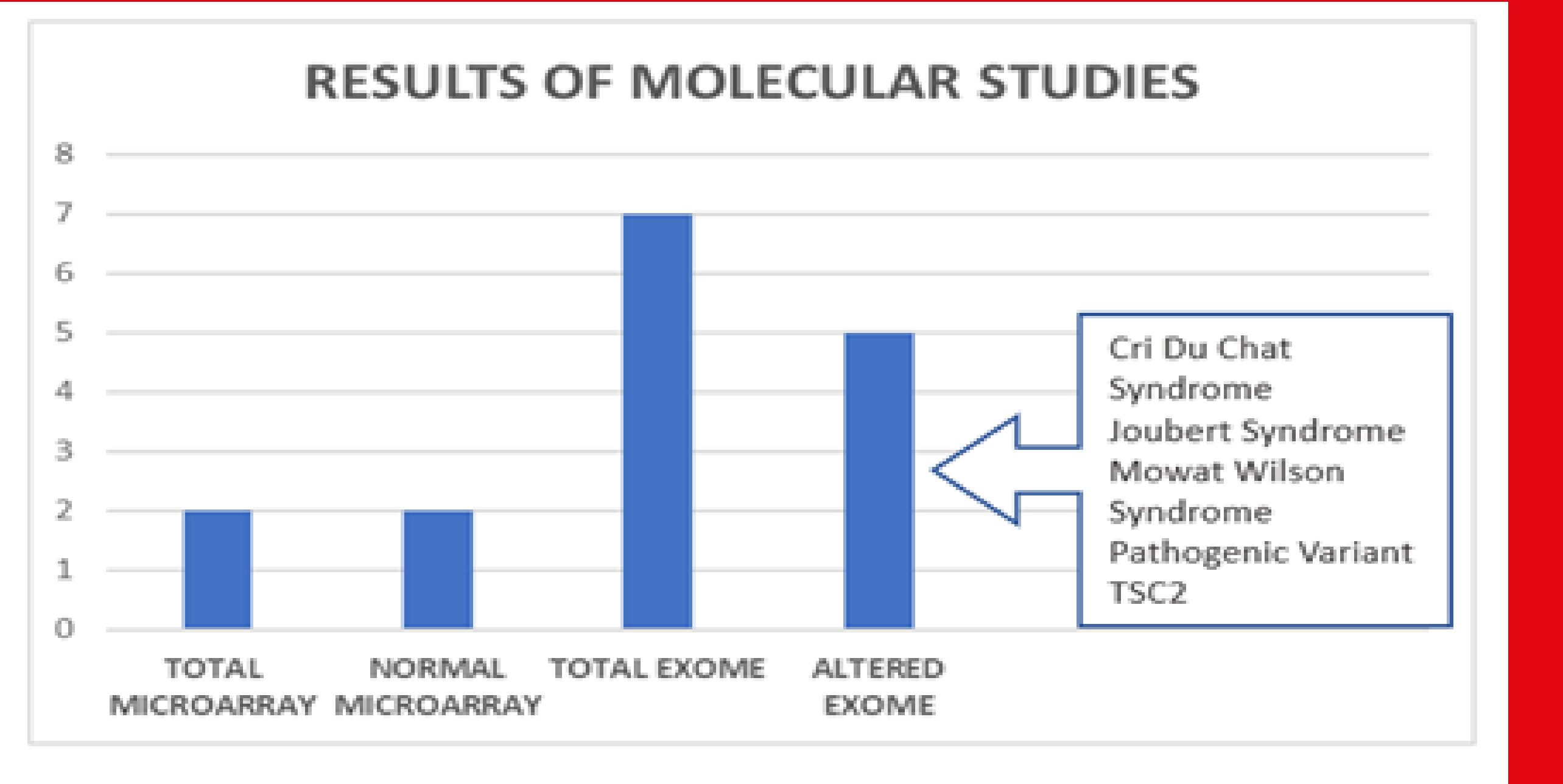


Figure 2. Results according to molecular genetic study