PSYCHOMOTOR DEVELOPMENT OF CHROMOSOME 13Q DELETION SYNDROME. A CROSS-SECTIONAL STUDY.

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INTRODUCTION

Chromosome 13q deletion is a rare genetic disease caused by the loss of genetic material from the long arm (q) of chromosome 13. It is characterized by clinical variability depending on the region of the chromosome affected.

THE PURPOSE OF THE STUDY

Analyze the pattern of psychomotor development in people with chromosome 13q deletion syndrome and prepare an informative document for family members and/or professionals that includes the most relevant clinical characteristics.

MATERIAL AND METHODS

Cross-sectional study through a self-prepared survey, due to the non-existence in the literature of a specific questionnaire on the characteristics and evolutionary development of this pathology or a similar one. The survey is intended for family members or caregivers of the person affected by the syndrome. The analysis of the results was carried out through the Excel program.

TARGET POPULATION

The sample consisted of 17 people with a diagnosis of deletion syndrome or partial monosomy of chromosome 13q and a different country of origin. The mean age was 13.39 (range 14 months to 42 years) and 59% were male.

For the study of some variables, especially motor variables such as crawling, walking, riding a bike, people younger than the age necessary to acquire a specific skill were excluded from the analysis.

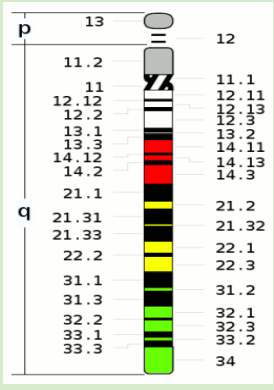
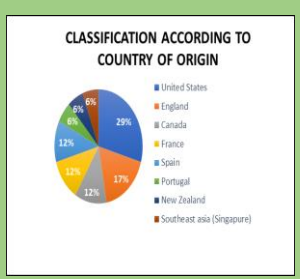


Image 1. Regions of chromosome 13. Source: Chromosome Disorder Outreach Inc. (CDO)

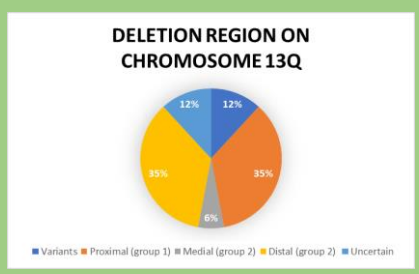
MEASURES, PROCEDURE AND STATISTICAL ANALYSIS

- For the recruitment of participants, contact was made with associations from several countries through different internet platforms.
- A survey based on the Alberta infant motor scale, Denver Scale and Haizea Llevant scales was developed, which collects data on genotype-phenotype relationship, clinical complications at birth, age of diagnosis, physical malformations and intellectual disability, management in activities of the daily life and the acquisition of psychomotor milestones corresponding to the developmental age.
- To analyze the variables, the correlation of the clinic with the affected region of chromosome 13q is studied.
- After analyzing the results of the survey, an informative document was prepared for families and professionals.
- The analysis of the results was carried out through the Excel program.

RESULTS



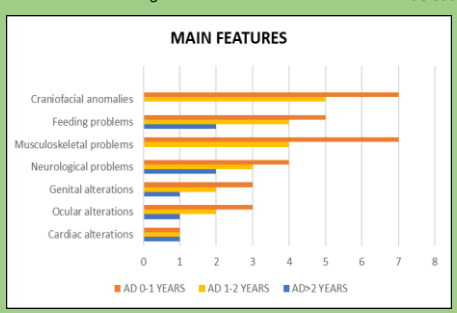
Graph 1: Classification of the sample according to the country of origin.



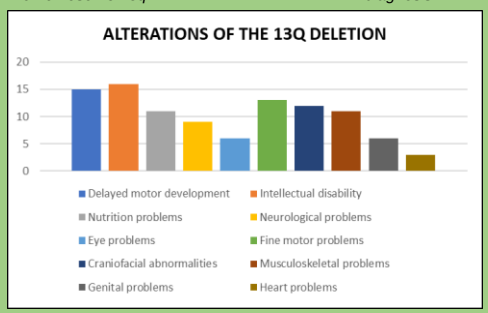
Graph 2: Classification of the sample (n=17) according to the region of deletion in chromosome 13q.

Graph 3: Classification of the sample according to age at diagnosis.

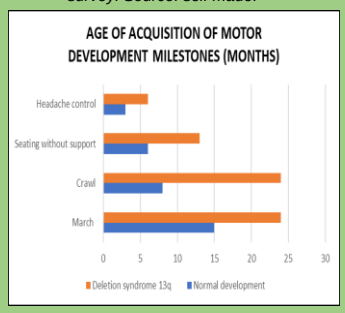
Image 2. Informative document with the information collected through the survey. Source: self made.



Graph 4: Relationship between the age at diagnosis and the clinical characteristics present in the sample (n=17).



Graph 5: Classification of the main alterations present in the sample (n=17).



Graph 6: Comparison between the age of acquisition (months) of motor milestones of the participants in the sample (n=17) and the values of normative development.

DISCUSSION

- The information reported through the survey coincides with that published in the medical literature, with the exception of heart and eye involvement (retinoblastoma) observed in different regions of chromosome 13q, not previously described.
- To date, there is no validated questionnaire in the literature that collects all the variables analyzed in this study for 13q deletion syndrome.

CONCLUSIONS

- ✓ The 13q deletion syndrome can be classified into four groups, almost half of the deletions are proximal or distal.
- ✓ The most severe phenotypes correspond to the less common variants and to the middle region. One of the most frequent alterations is intellectual disability and, therefore, impairment in communication and functional activities.
- ✓ Most of the respondents need help to perform basic activities of daily living.
- ✓ Most patients make use of rehabilitation from a very early age.
- ✓ The impediment that professionals can find is the scarcity of available information, in the field of physiotherapy there are no specific guidelines for the physical characteristics and the development of the pathology.

BIBLIOGRAPHY

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3. Cuadrado Martín M, Boldova Aguar C, Carrasco Lorente S, Martínez Laborda S, López-Pisón J, Baldellou Vázquez A, et al. Monosomía r (13): a propósito de una nueva observación. Anales de Pediatría. 2000;53(6):592–5.

QR1: Informative document with the information collected through the survey. Source: self made.

QR2: Complete bibliography of the study