Intellectual Disability Related to De Novo Germline Loss of the Distal End of the P-Arm of Chromosome 17: A Case Report

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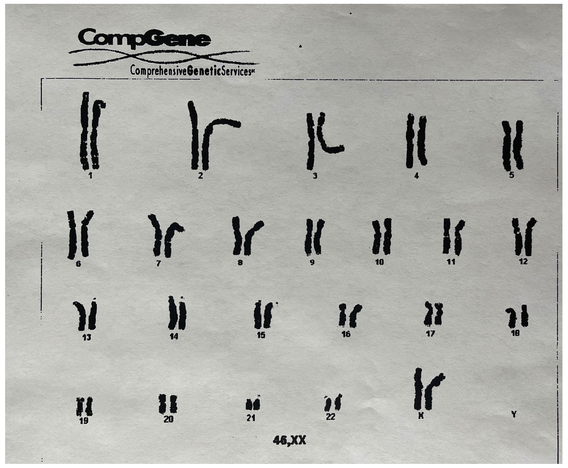
Background

In this report we present a case of a 20-year-old female with congenital intellectual disability, stunted growth, and hypothyroidism. Competitive genetic hybridization (CHG) revealed a loss of 17p13.3; the deletion was not present in either parent. Mutations on the p-arm of chromosome 17 are associated with Miller-Dieker Syndrome and Isolated Lissencephaly Sequence, both of which share symptoms in common with the patient.



Picture 1. Chromosome karyotyping when pt. Was born.

Methods

Peripheral mononuclear cells (PBMCs) were used for karyotyping and competitive genetic hybridization (CHG). Bioinformatic analysis was carried out using the Genome Data Viewer (ncbi.nlm.nih.gov/genome/gdv). Assent was obtained from the patient and consent was obtained from the patient's parents prior to beginning the study.

Results

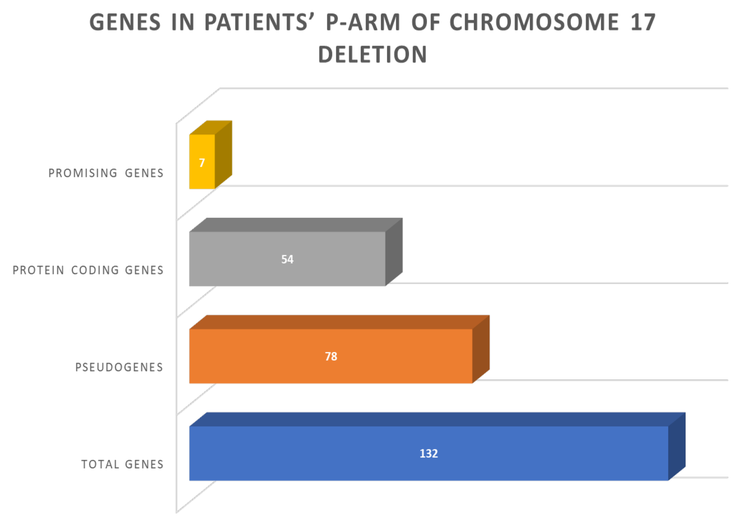


Figure 1. Each gene present in the P-arm of chromosome 17 was researched utilizing NCBI for functionality. The classifications of each of the coding regions present in this region were then identified for potential linkage to the patient's presentation.

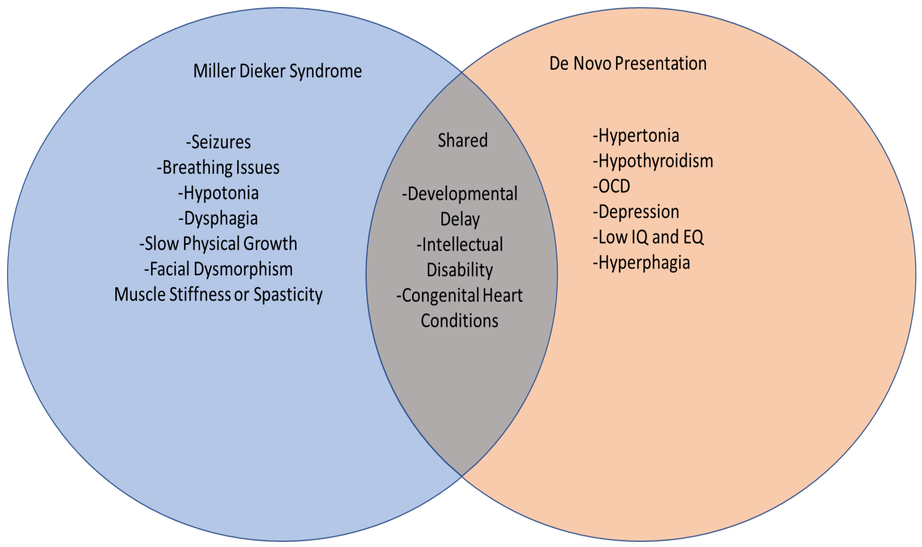


Figure 2. Upon analysis of the genes present in the P-arm of chromosome 17 Miller Dieker Syndrome became a differential based upon the karyotype. Herein is demonstrated the clinical presentation of the patient in comparison to classical Miller Dieker Syndrome.

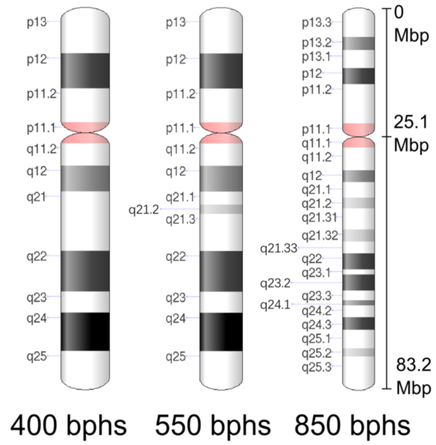
Results Continued

17p13.3 gene deletions associated with MDS:

- HIC1
- PAFAH1B1
- YWHAE

17p13.3 genes potentially related to patient's symptoms:

- DPH1
- CCDC92B
- RAP1GAP2
- TAX1BP1
- SMG6



Conclusion

The mutations of the short arm of chromosome 17p13.3 sequence, affect neurodevelopment and typically leads to intellectual disabilities, relatively short stature and a lack of empathy. This data provides additional details regarding intellectual disability related to de novo germline loss of chromosome 17 and possibly provide further insight in correct diagnosing and treatment options. However, further research and testing is needed.