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Search for the mutation causing the thoracoabdominal syndrome (TAS) an x-linked dominant disorder

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Introduction: The thoracoabdominal syndrome (TAS) is a very rare congenital x-linked dominant disorder presented only in one Jewish North African family. The features of the syndrome are ventral herniae (midline abdominal wall defect), antero-lateral diaphragmatic herniae (manifested almost exclusively in affected males), hypoplastic lung, and associated cardiac anomalies. The aim of this study is to identify and characterize the mutation causing TAS.

Materials & Methods: The initial search for the mutation causing the syndrome was done by linkage analysis on the x chromosome using 27 individuals of the TAS family. Genome sequencing of two of the family members was performed on the identified TAS interval on chromosome X.

Results: The TAS interval was localized to Chr:Xq27.1 in an interval of 1.06 Mb. Three genes and two miRNA are encoded in the interval. No mutation causing variants were identified in these sequences. Large deletions, insertions and translocations were negated by the sequence. Therefore, further search for indel variants in the non-coding DNA was pursued. Variations presenting in more than 1 person in the population databases and in short tandem repeats (STR) regions were excluded, leaving 4 variations that are presently being analyzed specifically for prevalence in the Jewish North African population.

Conclusion: Further analysis using the chromosome conformation capture-on-chip (4C-seq) technology will be held on the mutation causing variant for determining long-range chromatin interactions and the gene that may be affected.

Biography

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