When a child is diagnosed with a genetic condition, one of the first and most important questions is what to expect in terms of their health and development. In the case of distal 18q-, this is a particularly difficult question. Each person with distal 18q- has a different breakpoint. That means that different people are missing different genes. Therefore, it is very hard to create a general description of distal 18q-. However, there is a group of people who have different 18q breakpoints that are between the same two genes. This means that, as far as the genes are concerned, these people had the same deletion. The purpose of this paper was to establish a “reference group” of distal 18q-, with the understanding that there will be a great deal of variation in the people with distal 18q-. We reviewed the records of 16 people who had similar sized deletions that, despite different breakpoints, contain the same set of genes. Their deletion was about the average size when compared with other individuals in the study. Fourteen of these participants came to San Antonio for a series of clinical evaluations. The table below indicates the incidence of the most common findings in this group.

In terms of cognitive abilities, the majority of this study group performed within the low normal range. However, they had problems in adaptive abilities, or the skills needed to adjust to and function appropriately in day-to-day life.

This paper also reviewed the different genes that are located within the deletion. There are six in this region that may play a role in causing the different issues associated with distal 18q-. These genes are TNX3, NETO1, ZNF407, TSHZ1, NFATC, and ATP9B. These six genes aren’t responsible for all of the features of 18q-. However, the may contribute to some of the findings. As time goes on, our understanding of the roles of these genes will advance.