
One of the primary goals of the Research Center is to find the genes that cause the different features associated with conditions involving chromosome 18. The first step in this process is to narrow the search down to the regions of the chromosome where these genes are located. These are called “critical regions”. In this paper, we reported the critical regions for four well-known features of 18q-: kidney malformations; dysmyelination of the brain; growth hormone stimulation response failure; and aural atresia. All of these critical regions are located near the end of the long arm of chromosome 18 and overlap with each other significantly. People missing the critical regions have a 25% chance of a kidney malformation; a 100% chance of dysmyelination of the brain; a 90% chance of growth hormone stimulation failure; and a 78% chance of aural atresia. We still have a lot of work to do to identify the genes responsible for these conditions, but we’re definitely making progress! Also, this allows us to make some general predictions about what to expect for people whose deletions include these critical regions. In fact, this is discussed in greater depth in another manuscript we published in 2009, titled, “A gene dosage map of chromosome 18: A map with clinical utility”.