
It has been well-established that growth hormone deficiency is frequently identified in people with chromosome 18 changes. We wanted to determine which genes on chromosome 18 are responsible for the growth hormone deficiency in 18q- and 18p-. We collaborated with an Eli Lilly research program to see if we could identify people with very small chromosome 18 deletions that involve one or only a few genes. Eli Lilly had a collection of DNA samples from people with isolated growth hormone deficiency not caused by a known gene mutation. The same technology we use to study chromosome 18 changes also allows us to look for changes on other chromosomes. We looked at 45 people with isolated growth hormone deficiency. Although we did not find any people in this group with chromosome 18 changes, we did find three individuals with changes on other chromosomes that had not been previously reported. These chromosome regions are located at 13q33.1 and 20q13.13. Additional studies are necessary in order to identify the specific genes that play a role in growth hormone deficiency.