
In recent years, several genes have been identified on 18p that are associated with specific conditions. One of these genes, SMCHD1, has been linked with facioscapulohumeral muscular dystrophy, type 2 (FSHD2). This condition leads to a specific pattern of weakness mainly affecting the face, shoulder blades, upper arm, lower legs, and hips. It typically onsets during the teen years. The initial reports of SMCHD1 included people with point mutations in the gene that render the gene nonfunctional. A point mutation means that there is a change in one base pair in the gene. This is different from a deletion of the entire gene, which is what occurs in people with 18p-. This paper reported on two families that were diagnosed with FSHD2 who also had a small interstitial 18p deletion that included the SMCHD1 gene. However the members of these families with FSHD2 also had a variant on chromosome 4 where the primary gene for FSHD lies. Therefore, it appears that whole gene deletions of SMCHD1 can lead to the features of FSHD2 but only when the chromosome 4 variant is also present. Therefore, only a small minority of people with 18p- are actually at risk for FSHD2. Additional research is necessary to learn more about FSHD2 presents in people with 18p- as well as how it progresses.