
18p- was first described years ago. However, for the first several decades, description of this condition were limited. Technology limited the ability to determine the precise breakpoints. This paper was the first to describe a relatively large set of individuals with 18p- that share the same breakpoint. In this paper, we described 31 people with 18p- and a breakpoint at the centromere. The most common findings included neonatal complications; cardiac anomalies; hypotonia; MRI abnormalities; endocrine dysfunction; strabismus; ptosis; and refractive errors. Less common features included holoprosencephaly and its microforms; hearing loss; and orthopedic anomalies. We also reported on the cognitive status of people with 18p- as well as behavioral concerns. The average IQ score was in the borderline range of intellectual functioning. Although cognitive abilities were higher than reported in the older literature, there were some behavioral concerns. It appears that these individuals difficulties with activities of daily living. This paper is a first step in linking specific genes on 18p with the various features of 18p-.