
Abstract: The pericentromeric region on 16p appears to be susceptible to chromosomal rearrangements and several patients with rearrangements in this region have been described. We report on a further patient with a microdeletion 16p11.2-p12.2 in the context of described patients with a deletion in the pericentromeric region of 16p. Minor facial anomalies, feeding difficulties, significant delay in speech development, and recurrent ear infections are common symptoms of the microdeletion syndrome 16p11.2-p12.2. All reported patients so far share a common distal breakpoint at 16p12.2 but vary in the proximal breakpoint at 16p11.2. The microdeletion 16p11.2-p12.2 should be distinguished from the approximately 500 kb microdeletion in 16p11.2 which seems to be associated with autism but not with facial manifestations, feeding difficulties, or developmental delay.