CMA Case 1

A 3-year-old boy with achalasia and short stature

A 3-year-old boy developed recurrent vomiting during infancy and was diagnosed to have achalasia that required surgical intervention. He also had global developmental delay, failure to thrive, short stature, hypotonia, strabismus, decreased tearing, and distinctive facial features (cupped ears and retrognathia). Family history: Parents are first cousins and they and two brothers are healthy.

Regions of homozygosity **using hg19 assembly**

chr1:154,335,417-175,755,901
chr2:47,382,606-61,245,908)
chr2:174,007,883-226,827,297
chr4:59,970,766-65,510,854
chr4:71,290,636-77,773,437
chr5:133,423,890-141,765,951
chr7:15,669,000-50,811,561
chr7:83,700,685-122,712,358
chr8:22,606,613-41,573,488
chr10:10,586,371-30,254,648
chr10:94,492,716-113,507,841
chr12:95,118,069-98,234,300
chr18:7,448,001-11,564,311
chr22:29,491,514-39,183,883