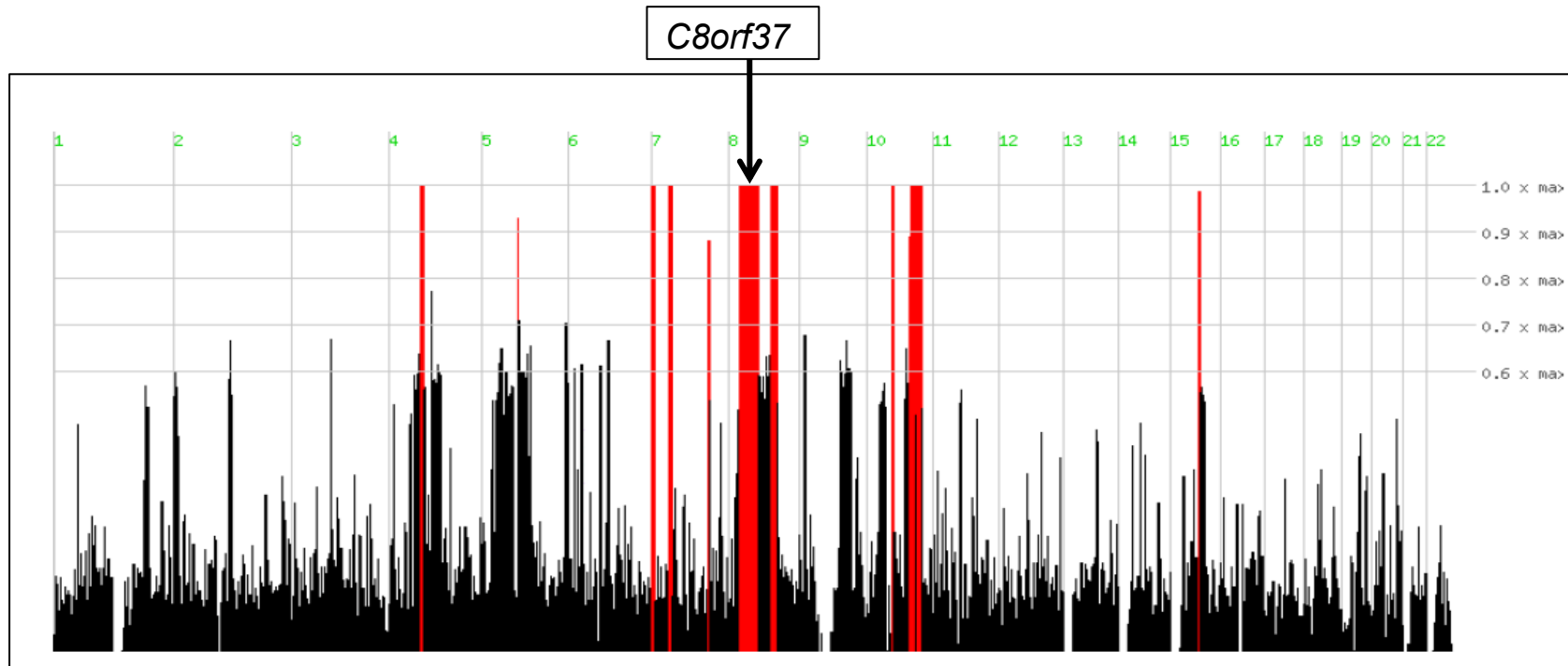


## Appendix 2. Homozygosity mapping in family MA48.



Analysis of the SNP genotyping data of affected cases 48-2, 48-3 and 48-7 identified autozygous segments on chromosomes 4 (rs10212780 – rs3755967, ~7.1Mb), 5 (rs4357016 – rs1443096, ~2.0Mb), 7 (rs6583338 – rs4724950, ~7.2Mb; rs4720204 – rs11771663, ~5.4Mb and rs6943120 – rs12706290, ~2.6Mb), 8 (rs4445224 – rs7842637, ~79.0Mb), 10 (rs6537586 – rs1973972, ~61.5Mb) & 15 (rs16940472 – rs8042228, ~2.4Mb) that are highlighted in red. The location of *C8orf37* which is at ~14.4Mb on chromosome 8 is depicted.