

Appendix 5. A homozygous point mutation within ECR-7 segregates with MOTA. Electropherograms of a point mutation found to segregate with the disease are shown for the four probands and the unaffected parents in ECR-7, a region identified by ECR Browser to be conserved from human, chimpanzee, rhesus macaque, cow, opossum, rat, mouse, chicken and frog, but not in puffer fish or zebrafish. Probands are homozygous C/C while unaffected parents are C/T. The genomic reference used to compare sequence is T/T in this position.”

