Unaffected parental cells are carriers of different mutations within the same gene.

Affected proband inherits different pathogenic mutations, one from each parent, giving rise to the compound heterozygote state. Both the maternally and paternally inherited genes are compromised by different mutations and the proband has no wild-type alleles.

Unaffected parental cells are carriers of the same mutation within the same gene.

Affected proband inherits the same pathogenic mutation, one from each parent, giving rise to the homozygote state. Both the maternally and paternally inherited genes are compromised by the same mutation and the proband has no wild-type alleles.

Following meiosis and recombination some parental gametes (egg and sperm) contain the mutation.

Maternal Paternal

Child

Autosomal recessive

Maternal Paternal

Child

Compound heterozygote

Chromosome

Mutation 1

Mutation 2