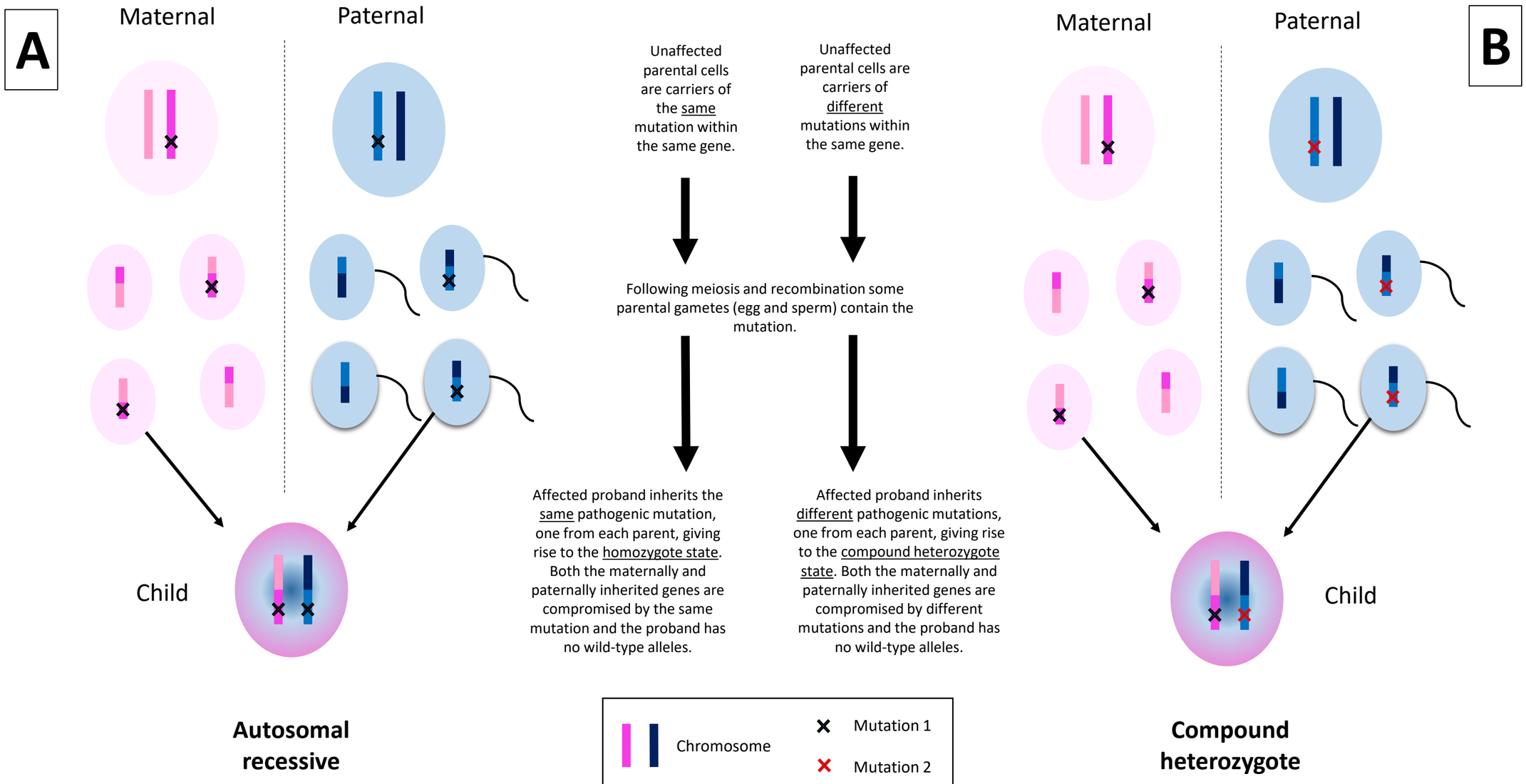
A
Maternal
Paternal
Unaffected parental cells are carriers of the same mutation within the same gene.
Unaffected parental cells are carriers of different mutations within the same gene.
Following meiosis and recombination some parental gametes (egg and sperm) contain the mutation.
Affected proband inherits 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.
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.
Child
Autosomal recessive
Chromosome
Mutation 1
Mutation 2
B
Maternal
Paternal
Child
Compound heterozygote