



General introduction

Recent progresses in research have clearly shown that some rare fever diseases are caused by a genetic anomaly. In many of them, other members of the family can also suffer from recurrent fevers.

What does genetic anomaly mean?

This means that a gene has been modified by an accident called a mutation. This mutation changes the function of the gene which gives wrong information to the body and results in the disease. In every cell, there are two copies of each gene. One copy is inherited from the mother and the other copy is inherited from the father. The mutation can be

a) present in the parents. The inheritance is of 2 different types:

-recessive: that means that both parents carry the mutation, on only one of their two genes. They are not ill because the disease occurs only if the two genes are affected. The risk for a child inheriting the mutation from each parent is one in four.

-Dominant: that means that one mutation is enough to express the disease. In that case, one of the parents is ill, and the risk for transmission to the child is one in two.

b) absent in the parents. The accident has occurred during the child's conception. It is called de novo mutation. There is theoretically no risk for another child (no more than random), but the affected child's offspring has the same risk of being affected as with the dominant mutation (i e one in two).