www.printo.it/pediatric-rheumatology/ZA_GB/intro

Autoinflammatory Diseases

Version of 2016

1. GENERAL INTRODUCTION TO AUTOINFLAMMATORY DISEASES

1.1 General introduction

Recent progress in research has clearly shown that some rare fever diseases are caused by genetic defects. In some of diseases there may be several members of a single family who suffer from recurrent fevers.

1.2 What does "genetic defect" mean?

A genetic defect describes a gene that has been changed by an event known as a ‘mutation’. These mutations can change how vital body processes work and give incorrect information to the body, resulting 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. Diseases can be inherited in different ways:
Recessive: in this case, both copies of the gene carry a mutation. Parents usually carry the mutation on only one of their two genes and they are not ill as the disease occurs only if both genes are affected. The risk for a child inheriting the mutation if both parents are carriers is one in four. Dominant: in this case, one mutation is enough to express the disease. If one of the parents has the disease, the risk for transmission to a child is one in two. Sometimes a child has a disease but neither of their parents carry the mutation; this case is known as ‘de novo mutation’. The accident affecting the gene has occurred at the child’s conception. In this case there is theoretically no risk for the child’s brothers and sisters (no more than random), but if the affected child goes on to have children the risk to them is one in two (the same

as for a dominant mutation).

1.3 What is the consequence of the genetic defect?

The mutation will affect its specific protein and alter how the protein does its job. The mutated protein will favour the inflammatory process and allow that triggers, which are not big enough to cause inflammation in healthy people, will induce fever and inflammation in the affected person.