

Title: Autoinflammatory diseases and genetic tests: friends not turning to foes

Autoinflammatory diseases are a group of genetically heterogeneous diseases. In the past, many of these diseases have been misdiagnosed therefore causing delay in starting effective treatment which can result in permanent organ damage e.g. intellectual disability, deafness and blindness or early onset renal failure secondary to amyloidosis. This not only affects the quality of life of the patients but also burdens the families heavily. With the advancement of new technologies in genetic tests, next generation sequencing (NGS) methods (targeted sequencing of a gene panel or whole exome sequencing) has allowed timely genetic characterization of many of the diseases at an affordable cost. However, this is not the ultimate solution and answer for autoinflammatory or any other inheritable diseases. There are on-going and heated discussions on genotype/phenotype correlations, role of environmental triggers of gene expression, the sequencing of non-exomic DNA consisting of introns and regulatory regions of a gene (whole genome sequencing), the dilemma of detecting a carrier state or sequences/variants with unknown clinical significance. Nonetheless, it is undeniable that genetic technologies are a powerful tool. In order to make the best use of this tool and benefit our patients and their families to the best extent, we have to apply the tests wisely and strategically. We can make the headway by promoting three things in our daily clinical practice: acquiring a good knowledge of this ever-expanding group of diseases through easily accessible learning platforms, arranging genetic counselling services to patients and families both before and after genetic tests and contributing to the established genetic database of autoinflammatory diseases.