Genotyping of MEFV and SAA1 Genes and Their Correlation to the AA-Amyloidosis Development

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Abstract:
Background: Familial Mediterranean fever (FMF) is the most common autoinflammatory disease caused by recessive mutations in the MEFV gene. If not treated, FMF patients may develop renal AA-amyloidosis that leads to renal failure and death. Both mutations and polymorphisms in MEFV and SAA1 genes, respectively, have been associated with AA-amyloidosis in several populations. In Algeria, as FMF is still under-estimate and misdiagnosed, genetic data on renal complication are largely lacking. We thus explored the contribution of MEFV and SAA1 loci in the development of amyloidosis in Algerian patients with FMF.

Methods: This study included 64 unrelated FMF patients (21 without and 43 with renal amyloidosis) and 13 healthy controls. The entire exon 10 was sequenced after PCR amplification to detect MEFV mutations. Genotypes of SAA1 locus (SAA1.1, SAA1.5, and SAA1.3) were determined by PCR-RFLP (restriction fragment length polymorphism).

Results: Analyze of MEFV gene showed that the percentage of homozygous for p.M694I mutation was significantly higher in patients with amyloidosis compared to patients without amyloidosis (p=0.032). The SAA1.1/1.1 genotype was significantly predominant in patients with amyloidosis compared to those without AA-amyloidosis (p=0.001) and controls (0.001). The SAA1.5/1.5 genotype was identified only in patients without amyloidosis and controls. The most patients with renal complications were homozygous for p.M694I and SAA1.1 alleles.

Conclusion: Our data suggest a positive correlation between the p.M694I/M694I and SAA1.1/1.1 genotypes and the development of AA-amyloidosis secondary to FMF in Algerian patients.

Keywords: AA-Amyloidosis, Familial Mediterranean Fever, MEFV Gene, SAA1 Polymorphisms.

1. Declaration of conflicts

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2. Authors’ Biography

No biography

3. References

No references