GENETIC OF CALCIUM KIDNEY STONES

GIUSEPPE VEZZOLI[1]*, TERESA ARCIDIACONO[1], FRANCESCO RAINONE[1], ANNALISA TERRANEGRA[2], ELENA DOGLIOTTI[2], ANDREA ALOIA[2], LAURA SOLDATI[2]

[1]*San Raffaele Scientific Institute, Nephrology Unit, Milan, 20132, Italy*
[2]*Department of Medicine Surgery and Dentistry, Milan, 20142, Italy*
vezzoli.giuseppe@hsr.it

Calcium nephrolithiasis is one of the most prevalent uronephrologic disorders in the western countries. Studies in families and twins evidenced a genetic predisposition to calcium nephrolithiasis.
Family-based or case–control studies of single-candidate genes evidenced the possible involvement of calcium-sensing receptor (CASR), vitamin D receptor (VDR), and osteopontin (OPN) gene polymorphisms (SNPs) in stone formation.
The only high-throughput genome-wide association study identified claudin 14 (CLDN14) gene as a possible major gene of nephrolithiasis.
Specific phenotypes were related with these genes: normocitraturia with CASR gene SNPs, hypocitraturia and severe clinical course with VDR gene SNPs, and hypercalciura with CLDN14 gene SNPs.
The pathogenetic weight of these genes remains unclear, but an alteration of their expression may occur in stone formers.
Technological advances and accurate clinical examination may get new insight about the genetic basis of nephrolithiasis.

Keywords: nephrolithiasis, hydrophilic clay-affinitive