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Molecular Genetic Testing in Cystinuria

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ABSTRACT Cystinuria (OMIM 220100) is caused by the defective transport of cystine and the dibasic amino acids in the proximal renal tubule and in the epithelial cells of the gastrointestinal tract. We analysed a cohort of 26 unrelated cystinuria patients diagnosed on the basis of stone formation. Direct sequencing of all coding regions and exon-intron boundaries of the *SLC3A1* and *SLC7A9* genes allowed us to identify 26 different mutations in 23 out of the 26 patients, in total they accounted for 40 affected chromosomes. Three of the 26 are novel mutations, two in *SLC3A1* and one in *SLC7A9*. Interestingly, two of our patients carried three mutations in *SLC3A1* each, one patient was mixed heterozygous for *SLC3A1* and *SLC7A9* mutations. In summary, these findings expand the spectrum of *SLC3A1* and *SLC7A9* mutations and confirm the heterogeneity and complexity of cystinuria. If we assume an autosomal recessive inheritance of the disease, our detection rate was 88.5% and thereby relatively high in comparison to other studies. Nevertheless we have to consider that at least *SLC7A9* mutations are often dominant, we therefore think that our effective detection rate is higher. Additionally, the broad pathophysiological consequences of *SLC7A9* mutations make an individual prognosis and genetic counselling difficult.