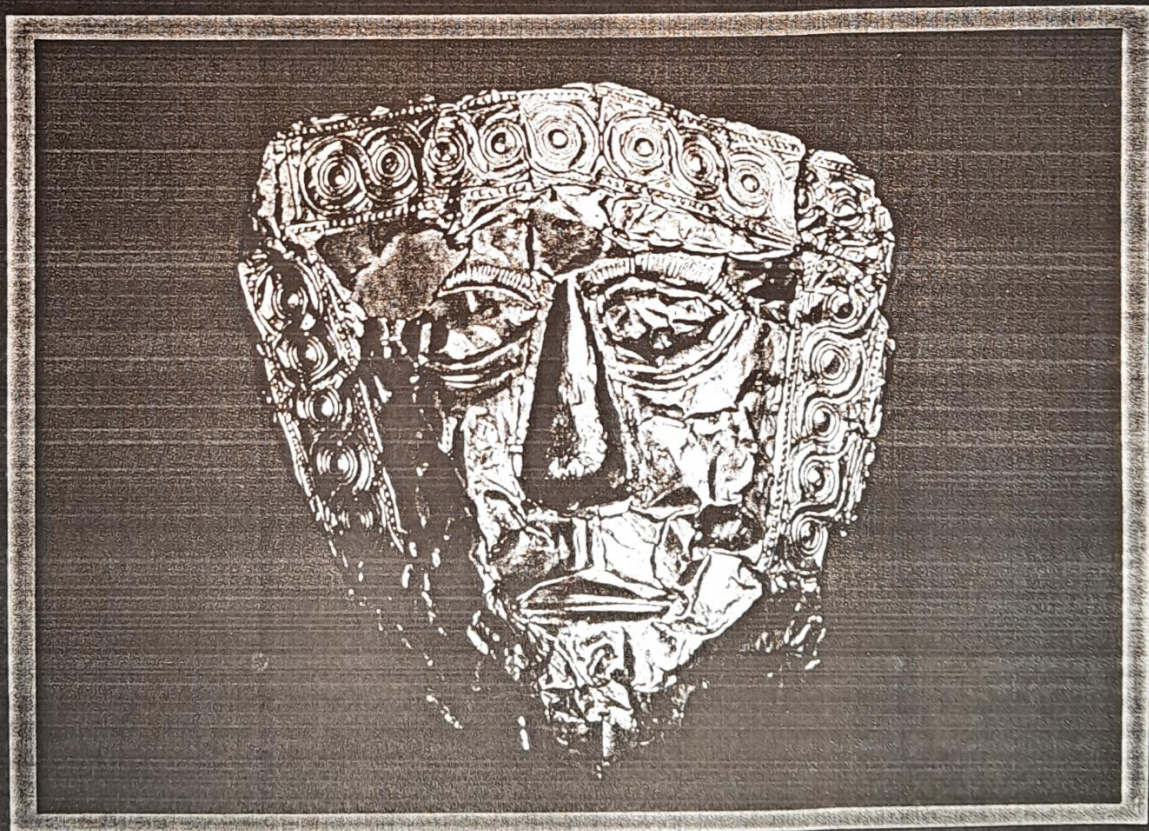


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**PP174. A new familial mutation in the SRY gene (Arg133Gly)**

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Mutations in the testis-determining gene SRY result in XY sex reversal with pure gonadal dysgenesis (PGD). Most of the SRY mutations affect the HMG-domain of SRY which plays a central role in DNA binding and bending activity of SRY. The amino acid codon 133 is conserved in the SRY gene of all mammal species. It is part of the basic C-terminal region of SRY, which was proposed to provide nuclear localization and transcriptional activation. A de novo Arg133Trp mutation was detected in two unrelated patients with pure gonadal dysgenesis affecting Arg133. Here we describe a similar mutation that affects codon 133 of the SRY gene, resulting in an arginine to glycine substitution in the protein. It was detected in a patient with PGD, who is a phenotypically normal male. However, the mutation was not found in the SRY gene of 150 other males, thus excluding the possibility of a common polymorphism. Our report of familial Arg133Gly mutation suggests that replacement of Arg 133 of the SRY is not sufficient for impaired organogenesis and emphasizes the importance of modifier genes in the sex determination pathway.

**Key words:** SRY, XY females, pure gonadal dysgenesis, familial mutation

**PP175. Five novel mutations in cystinuria genes SLC3A1 and SLC7A9**

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Cystinuria is an autosomal recessive disorder that is characterized by an impaired transport of cystine and dibasic amino acids in the proximal renal tubule and epithelial cells of the gastrointestinal tract. This defect permits an elevated urine concentration of cystine, which combines and precipitates as cystine stones. The transport is mediated by the SLC3A1/SLC7A9 transporter, the subunits of which are encoded by the SLC3A1 and SLC7A9 genes.

Cystinuria is classified into two types: type I and non-type I. Mutations in type I cystinuria, while responsible for the present two novel mutations (G242R and L573A) and three in SLC7A9 gene (G73K-V375I, L231-1236delACTC). The mutations were determined by direct sequencing. G242R and L573A mutations in SLC3A1 gene, and G73K mutation in SLC7A9 gene were found in patients from Serbia, V375I in SLC7A9 gene was found in a patient from Macedonia, while L231-1236del ACTC mutation in SLC7A9 gene was found in a patient from Turkey.

**Key words:** Cystinuria, SLC3A1, SLC7A9, missense, deletion

## Proceeding

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