

## Short Communication

# Mutations of cystic fibrosis transmembrane conductance regulator gene in patients with Mayer Rokitansky Kuster Hauser syndrome

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## Abstract

**Background and Objective:** Mayer Rokitansky Kuster Hauser (MRKH) syndrome is characterized by Mullerian duct aplasia in an XX individual with female phenotype presenting primary amenorrhea at adolescence. This study was done to determine the mutations of cystic fibrosis transmembrane conductance regulator (CFTR) gene including  $\Delta F508$ , G542X, N1303K, W1282X in patients with MRKH syndrome.

**Methods:** This case-control study was performed on 25 females with MRKH syndrome and 25 healthy females. Blood sample was taken from each subject. DNA genomic was isolated by standard methods and common mutations of CFTR gene analyzed by ARMS-PCR.

**Results:**  $\Delta F508$  gene was found in 3 in case and one individual in control group. G542X, N1303K and W1282X gene was not detected.

**Conclusion:**  $\Delta F508$  gene was found in 12% of patients with MRKH syndrome.

**Keywords:** MRKH syndrome, CFTR gene,  $\Delta F508$  gene, ARMS-PCR

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