associated with filamentous structures and cytoskeleton formation. *SEPT9* plays a role in multiple cancers as either an oncogene or a tumor suppressor gene. DNA methylation of *SEPT9* gene is often observed in a wide variety of cancers in fact, hypermethylation of the gene was recently introduced as a biomarker in some cancers. But methylation of *SEPT9* and its effect on protein expression is unclear in bladder cancer. Therefore, aim of this study was to determine firstly *SEPT9* DNA methylation profiles and secondly the effect of its methylation on Septin 9 protein expressions in bladder cancer. Methylation pattern of *SEPT9* gene was analyzed by pyrosequencing. Protein level of Septin 9 was determinated by western blot in 40 bladder tumors relative to 5 normal bladder controls. Sequencing analysis revealed significantly lower methylation frequencies in bladder tumor samples relative to normal samples ($p<0.01$). On the contrary, an increase in protein level was observed in almost half of samples. The reason for this condition may be post transcriptional regulation. As a result, *SEPT9* may be playing a role as oncogene and suggesting a need for further investigation on the explanation of oncogenic role on bladder cancers. Hypomethylation of *SEPT9* can be used novel candidate prognostic markers for bladder cancer.

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Our experience in BRCA-associated hereditary breast and ovarian cancer syndrome

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BRCA1- and *BRCA2*-associated hereditary breast and ovarian cancer syndrome (HBOC) is characterized by an increased risk for mainly female and male breast cancer and ovarian cancer. 80% of pathogenic variants in *BRCA1* and *BRCA2* are detected by sequence analysis and approximately 10% by deletion/duplication analysis. In our center, the results obtained by studying the whole gene sequence analysis of *BRCA1* and 2 with NGS (Next Generation Sequencing) method were compiled. Pathogenic variant was detected in 5 of 30 patients and VUS was detected in 2 patients. Four of the pathogenic variants are located in *BRCA1*; one was detected in *BRCA2*. Three of the pathogenic variants were frameshift (*BRCA1*:c.2215_2216insCT, *BRCA1*:c.2131_2132delAA, *BRCA1*:c.2952delT) and 2 were nonsense mutations (*BRCA1*: c.1059G>A and *BRCA2*:c.469A>T). Also, *BRCA1*:c.9976A>T variant which causes stop codon formation is classified as benign. *BRCA1* and *BRCA2* mutations were detected in 16.6% of patients with breast and ovarian cancer. This frequency can be considered slightly higher compared to the literature.

OP-21-008

Targeted gene panel sequencing for hereditary kidney diseases: Efficiently detects candidate pathogenic variants related with these disorders

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Hereditary kidney diseases like Polycystic Kidney Disease (PKD) is a heterogenous form kidney disease which multiple genes have been found to be implicated in disease etiopathogenesis. Genetic diagnosis is highly important for hereditary kidney diseases in order to ensure the etiology of the condition and counsel the patients properly. Twenty-one patients who admitted to our clinic with PKD and/or other nephropathy syndromes were screened for 44 genes related with kidney disorders. The exons/exon-intron boundaries were amplified by using Nephropathies Solution kit in MiSeq instrument. Sophia DDM platform and Sophia Genetics' MOKA were used in variant analysis and annotation. Variant Classification was performed according to HGMD Professional, CentoMd and other available in-silico tools and ACMG variant pathogenicity classification. Several pathogenic (P)\likely pathogenic (LP)\VUS variants were found in 32 genes included in our gene panel. Twelve cases had 16 novel variants in the *CLCNKB*, *DSTYK*, *PKD1*, *PKD2*, *PKHD1*, *SLC12A1*, *SLC4A1* and *TTC21B* genes. Six cases had 5 P variants in PKD1 and CYP24A1 gene and three cases had a LP variant in the BSND and PKD1 genes. One frameshift novel deletion was identified in a case whose family and her previous generation were affected with PKD. In addition, 6 cases had also CNVs in *HNF1B*, *CEP290*, *CLCNKB*, *PKHD1* genes. Our study affirms that genetic screening of patients with PKD and other nephropathy syndromes by using targeted gene panel not only eases the diagnosis but also help counseling the patients and their family members for the risk of developing the disease.

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