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Combination treatment of usnic acid and sorafenib on hepatocellular carcinoma

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Sorafenib (SOR) is the only drug approved for the treatment of hepatocellular carcinoma (HCC). However, SOR can cause significant toxicity in patients and thus, it is necessary to develop new treatment strategies directed at toxicity reduction and higher efficacy. For this purpose, for the first time we investigated combined therapeutic effect of usnic acid (UA), a lichen metabolite, and SOR. SOR and UA were treated with UA (12.5 μM and 25 μM) and SOR (0.1 and 0.5 μM) for 12, 24 and 48 hours, and cytotoxic effects were determined by WST-1 assay in HepG2 and SNU-449 HCC cell lines. Furthermore, we performed Annexin V and cell cycle analysis to investigate the apoptotic effects of the combination treatment. As a result, the cell viability of the HepG2 and SNU-449 cells significantly reduced to 26.0% and 18.0% at in combination treatment 25 μM UA+0.5 μM SOR, respectively for 48 h ($p<0.01$). The combination treatment induced more apoptotic cell death in HCC cells than SOR and UA alone. We found that the percentage of total apoptotic cells increased to 75.9% and 83.4% in HepG2, SNU-449, respectively at 25 μM UA+0.5 μM SOR. Additionally, combination treatment of 25 μM UA+0.5 μM SOR induced a considerable increase from 38.7% and 65.6% in control cells to 92.0% and 73.2% in G0/G1 phase for HepG2 and SNU-449 cells, respectively. In conclusion, combine treatment with SOR and UA exhibited a strong synergistic interaction (CI<1) and inhibited HCC cell proliferation by apoptotic death.

OP-21-010

The role of NGS method in the diagnosis of periodic fever syndrome

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Periodic fever syndromes are a group of autoinflammatory diseases and characterised by recurrent episodes of fever and systemic inflammation occur in the absence of autoantibody production or identifiable infection. The most responsible genes in this group are MEFV, MVK, NLRP3 and TNFRSF1A. We examined the variants in aforementioned genes detected in 30 patients with NGS (next-generation sequencing). We were able to explain the clinic of 11 patients with MEFV analysis. Although pathogenic variants were detected in the MVK gene in two patients, it did not support the clinic due to its heterozygous form. 3 likely pathogenic variants and one VUS (variant of unknown significance) were detected in the NLRP3 gene. In the TNFRS1A gene, 2 likely pathogenic and 4 VUS were detected. As a result, with these results, we aimed to emphasize the importance of NGS method in the diagnosis of periodic fever syndrome.

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A case of early onset breast cancer with pathogenic variation in *STK11* gene

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Peutz-Jeghers Syndrome (PJS) is an inherited autosomal dominant disorder consisting of characteristic gastrointestinal (GI) hamartomas, mucocutaneous pigmentations and predisposition to GI, breast and other cancers. In this study, we aimed to present a common mutation in *STK11* gene in PJS case with early onset breast cancer. Our case is a 34-year-old woman. She was referred us because of the early onset recurrent breast cancer by oncology clinic. Pathology results were invasive ductal carcinoma and in situ multifocal solid papillary carcinoma on left breast. After adjuvant chemotherapy, hamartomatous polyp was detected in her colonoscopy following rectal bleeding. She had a history of left oophorectomy for benign reasons. We detected pigmentations on lower lip mucosa, a brown papule on nose and a brown macula at left palmar region. In her family history, it was learned that her mother was operated nasal polyp excision three times and aunt's daughter had a 45-year-old breast cancer diagnosis and her 28-year-old sisters had black spots on lips developed in childhood. Similar skin lesions were present when the patient was a child. Next generation sequencing analysis for Hereditary Cancer Risk Panel was studied for PJS genetic diagnosis. It detected nonsense, heterozygous, c.250A>T pathogenic variation (rs137853076) on the exon 1 of *STK11* gene (ENST00000326873). In conclusion, physical examination is indispensable for the diagnosis of hereditary cancer syndromes. Molecular diagnosis of hereditary cancer syndromes give patient's relatives the opportunity to early diagnose and also risk reducing measures can prevent cancer formation.

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