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Genetic testing and rare diseases in Europe: activities of the European Society of Human Genetics

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The European Society of Human Genetics (www.eshg.org; ESHG) is a non-profit, non-governmental organization which has two main aims: (i) to promote research in basic and applied human and medical genetics and (b) to ensure high professional standards in diagnostic and clinical practice. ESHG also facilitates contacts between scientists and professionals who share these aims, in particular those working and/or residing in Europe. ESHG was established in 1967 and is one of the founding members of the International Federation of Human Genetics Societies. ESHG also has several important committees such as the Annual Meetings Committee who selects venues for ESHG conferences (which have over 2000 participants, including an increasing number of exhibitors), the Scientific Programme Committee who prepares attractive scientific programs for these meetings, Educational Committee dealing with pre-graduate and post-graduate education in genetics, or the Public and Professional Policy Committee who issues consensual policy documents and/or position statements on current topics. Activities of its Quality Committee, dealing with coordination of external quality assessment in molecular genetics and cytogenetics stem from the European Commission Network of Excellence project EuroGentest (www.eurogentest.org) that aimed at harmonization and standardization of genetic services in Europe. ESHG works closely with European National Human Genetics Societies. Finally, since 80% of rare diseases are of genetic origin the activities of the Orphanet portal (www.orpha.net) in terms of genetic testing will be presented.

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Analysis of specific genes expression in intestinal ischemia-reperfusion injury in rats

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The ischemia-reperfusion of the small intestine induces an inflammatory response triggered by tissue injury which involves the action of cytokines and other inflammatory mediators. The accumulation of inflammatory mediators can increase bacterial translocation by damaging junctions between epithelial cells in the mucosa of the small intestine. The delicate balance of the cytokine expression seems to be a key factor which can lead to hyperinflammation or immunosuppression and development of Multiple organ dysfunction syndromes (MODS). The aim of this study was to analyse mRNA expression of specific apoptotic genes (Bcl2, Bax) and cytokines (TNF alpha, TGFB2, IL10, IL6 and IL1beta) in ischemia-reperfusion injury of the small intestine. In the experiment, male Westar rats underwent 1h ischemia that was performed by complete occlusion of mesenteric artery. Samples were harvested after 1 hour, 24 hour and 30 days of reperfusion. Total RNA was isolated from the complete wall of the small intestine and purified. mRNA quantification of specific genes was performed by real-time RT-PCR system Mx3000P using relative standard curve method. All the examined genes showed similar tendency. The quantity of mRNA reached the highest level after 1hour of reperfusion, and then (after 24 hours and 30 days of reperfusion) the mRNA level decreased significantly. After 24 hours of reperfusion, regeneration mechanisms in the small intestine have started. Ischemia-reperfusion injury is a complex and specific process. We can conclude that analysed cytokines are the main mediators with established correlation with trauma, and seem to be the target of research for the future therapeutic alternatives.

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Keywords: ischemia-reperfusion, apoptosis, real-time RT-PCR, cytokines, bax, bcl2, rat.

P13-82
Proteomic investigation of plasma membrane proteins involved in HBV infection

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Hepatitis B virus (HBV) is an enveloped DNA virus member of the *Hepadnaviridae* family. Infection with HBV is a very serious health problem, resulting in acute and chronic hepatitis, cirrhosis and frequently hepatocellular carcinoma. Although there is an efficient vaccine available, more than 350 million people are known to carry the virus around the world. The viral particle is composed of a nucleocapsid containg the partially double-stranded DNA genome surrounded by the viral envelope. Usually viral infection begins with receptor recognition at the host cell plasma membrane, followed by highly specific cell-virus interactions. The early steps of HBV entry in target cells are largely unknown, because of the poor infectivity *in vitro* and the absence of a robust tissue-culture model to support virus infection. In this study we have made use of the HepaRG cells, the only proliferating cell line permissive for HBV infection following a differentiation treatment, to identify proteins with potential role in the HBV life-cycle. Mass spectrometry analysis in differentiated and non-differentiated cells showed an increased expression of several proteins upon differentiation process. Of these targets, 5 proteins where further taken into consideration for validation by Western-Blot and Immunofluorescence. Consistent with the mass spectrometry data, an important up-regulation of 3 proteins was confirmed. Further studies, linking these proteins with the infection process of HBV will be performed.

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