21ème Conférence Scientifique Internationale

Horizon 2020

Avancées scientifiques et technologiques

15-16-17 avril 2015
Campus des sciences humaines
Campus des sciences médicales
Université Saint-Joseph de Beyrouth
Rue de Damas – Beyrouth

RÉSUMÉS

Organisée par
L’Association Libanaise pour l’Avancement des Sciences
et l’Université Saint-Joseph de Beyrouth
en collaboration avec
le Conseil National de la Recherche Scientifique au Liban
10% FBS, 1% P/S and 1% NEAA, and incubated at 37° with 5% CO2. The proliferation and viability test were performed using tetrazolium salt. Cells apoptosis was determined by Annexin V-Fluorescein isothiocyanate assay. The concentration of APP and caspase-3 were measured using ELISA kits. The assessment of APP mRNA expression was done using RT-PCR technique. Cells migration ability was evaluated using boyden chamber assay in 24-well plate. The effect of costunolide on APP production in vivo was tested in NSG mice. Results and discussion: Costunolide and BIBR1532 (5 or 10μM), two telomerase inhibitors, decreased significantly the APP production in cells supernatant and at the mRNA level, not by an apoptotic effect, but by their action on telomerase. This has been demonstrated by a siRNA test and an Annexin V affinity assay.

Interestingly, inhibition of PI3K/Akt/mTOR signaling pathway by wortmannin, PI322, GSK1056915 and rapamycin lead to a decreasing in APP concentration suggesting the involvement of this pathway in APP regulation. Moreover, an additive/synergistic effect was shown, when costunolide and BIBR were combined with the previous inhibitors. Decreasing in APP concentration was also shown when cells were treated SPC2013, a PKC inhibitor. Indeed, treated cells with IL-6 (25ng/ml) showed a transitional increasing of APP production after 8h and a decreasing effect after 10h, suggesting the involvement of IL-6 in APP production modulation. The migration ability of HepG2/C3A cells decreased significantly after treatment for 24h with costunolide at 10μM (p=0.0021), BIBR at 10μM (p=0.0043) and rapamycin, an mTOR inhibitor, at 200nM (p=0.0097). Costunolide administrated at 30mg/kg daily from day 12 of injection of HepG2/C3A cells, was found to decrease significantly the APP concentration in serum NSG mice compared with the vehicle control (p=0.0123). Conclusion: Both inhibition of telomerase and PI3K/Akt/mTOR signaling pathway decreased the APP production and the migration of HepG2/C3A cells.

Inborn Errors of Metabolism Highlights in Oman: Collaborative model
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Inborn errors of metabolism (IEM) are significant cause of morbidity and death among children. Remarkable emerging technology tandem mass spectrometry (MS/MS) allows the ability to increase drastically the number of IEM diseases that can be detected from a single dried blood spot. Developing countries face challenges in IEM analytical platforms and human resources. The USJ-newborn screening laboratory acquired MS/MS in 2007 and became a regional reference; represent the MENA region at International Society for Neonatal Screening Council (www.sinas-neoscreening.org), aim to transfer the knowledge to the region for many countries Iraq, Oman, Libya, Tunis and others. We are going to present the outcome of the collaboration with Royal hospital in Oman via Cerba-Paris. Among 516 samples were sent to USJ for IEM investigation in three years, 145 found to be positive cases. The high incidence is due that some were sick babies with symptoms evoking metabolic disorder: perinatal asphyxia, seizures, hypotonia, organ failure, psychomotor delay, mental retardation and others or with family history. Major finding were related to G6PD deficiency (117) and galactosemia (GAL) 8 cases. The etiology of these finding was supported by local physicians. The high incidence of G6PD is due to malaria resistance and there is a cluster of GAL in some tributes. This collaboration should continue meanwhile to assure know-how and take advantage transfer even after their inquiry of the equipment to reduce false negative and false positive rate as a case of pyruvate carboxylase was diagnosed as citrullinemia by their local resources.

Superior Mesenteric artery syndrome: clinical, endoscopic and radiological correlations according to a single-center experience
Giovanni Tomasello (University of Palermo, Italy)
Background and aim: The superior mesenteric artery SMA syndrome is a rare entity presenting with upper gastrointestinal tract obstruction and weight loss, due to the compression of the third part of duodenum between the aorta and the SMA. Studies to determine the optimal methods of diagnosis and treatment are essential. This study aims to analyze the clinical presentation, the diagnosis and the management of this syndrome. Material and methods: Over a 2-year period (2013-2014), 8 cases of SMA syndrome (out of 2076 gastroenterological endoscopies, EGDS)). Therefore, these patients performed computed tomography(CT) scan to confirm the diagnosis. Once the diagnosis was confirmed, the patients were referred to a gastroenterologist and to a nutritionist to discuss a personalized approach of therapy; furthermore, for each patient a surgical consultation was proposed. Results: In our series we evaluated retrospectively 8 cases of SMA 6 females), with a prevalence of 0.004%. Median age was 23.5 years (range 14-40), and median weight was 47.5 kilos (range 40-84). The median body mass index was 21 kilos/m2. Symptoms developed between 6 to 24 months (median 12). Premorbid conditions were present in four patients (Anorexia nervosa in two patients, and Spina bifida and Crohn’s disease in two patients). Only 2 of 8 patients were hospitalized, due to severe malnutrition. Median aorta-mesenteric angle was 22°, and median aorta-SMA distance was 5 mm. Interestingly, all the patients improved on conservative treatment. Conclusions: To date, SMA syndrome represents a diagnostic and therapeutic challenge. With regard to previous series published, our results show: the importance of the endoscopic suspicion of SMA syndrome, confirmed by CT scan; the preponderance of a longstanding and chronic onset; a female preponderance; the importance of the nutritional counselling in the therapeutic approach; the absence of need for surgical intervention; the better diagnostic accuracy of the narrowing of the aorta-SMA distance, rather than the narrowing of the aortomesenteric angle. Further prospective studies, with a larger number of patients, are needed to clarify the best way to diagnose and manage the SMA syndrome. 