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colocalized in mitochondria. We proposed that hMCM and hMMAA interact with each other by their cofactor domains.

097 - Population-Based Study of New Mutations Causing Sandhoff Disease in Argentina

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Background: Sandhoff Disease (SD) is a lysosomal storage disorder caused by mutations in the HEXB gene. A high incidence of SD has been described in an Argentine region called "Valle de Traslasierra." Mutations c.445+1G>A and p.S261Cfs12X were found in 98.7% and 1.3% of mutant alleles, respectively. In previous population-based studies, the carrier frequency has been estimated to be 1 in 16 to 29, all heterozygous with c.445+1G>A. Recently, we detected new mutations in 5 Argentinian patients: c.1082+5G>A, c.1242+1G>A, c.1451G>A (p.Gly484Glu), c.1597C>T (p.Arg533Cys) and c.1601G>A (p.Cys534Tyr). **Objective:** To study the heterozygote frequency for new mutations in the population at risk. **Material and Methods:** Blood samples were obtained from 200 healthy patients born in the region. Mutation analysis was performed by polymerase chain reaction/sequencing of specific DNA sequences. **Results:** We found 9 carriers of c.445+1G>A and none of other mutations among healthy patients (c.1082+5G>A has not yet been investigated). **Conclusion:** These results suggest that the frequency for new mutations is very low and confirm the role of c.445+1G>A as a founder mutation in the population at risk.

098 - Positive Pressure in the Treatment of Signs and Symptoms of Alveolar Hypoventilation and Sleep Disorders in Mucopolysaccharidosis

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Introduction: Mucopolysaccharidosis Home Care and Assistance Program (PMPS) of Children's Hospital John Paul II (CHJPII)/Hospital Foundation of the State of Minas Gerais (FHEMIG) provide assistance at home, and during outpatient admissions, to patients with MPS with cardiorespiratory involvement (CCR). As CCR worsens (decrease in vital capacity), it is evident in alveolar hypoventilation (HA) and sleep disorders (SD), first at night and the during the day. Cardiorespiratory signs and symptoms (SS) suggest AH and SD mainly dyspnea, airway hypersecretion, pulmonary hypertension, heart failure, difficult compensation, and intolerance to exercise. Treatment for these SS include establishment of positive pressure (PP) maneuvers through the air stacking (MAS) with a manual bag and mechanical ventilation (MV) performed regularly. **Objective:** To describe the results of the institution of PP in the treatment of hypertension in patients with DS and MPS accompanied by PMPS Program/CHJPII/FHEMIG. **Patients and Methods:** A quantitative, descriptive, observational, and retrospective study was held at CHJPII/FHEMIG/SES. Data were collected between 2007 and 2013. All patients accompanied by the Programs were included. **Results:** A total of 44 patients were followed (MPS types: 9 I, 9 II, 3 IIIa, 1 IIIb, 2 IVa, and 20 VI). Seventeen patients were diagnosed with AH and SD. Seventeen VM home users, perform 8 MEA. Median follow-up in Programs: 4.5 years. The SS shown before and after administration of PP: no patients (17×0), number of SD (median), (10×0), use of medications for treating SS (no patients; 17×4), hospitalization for SS (no patients; 9×0), deaths from SS (17×2). **Conclusion:** PP contributes to HA and SD treatment of SS patients with MPS.

099 - Preliminary Findings Evaluating Safety and Efficacy of Recombinant Human N-Acetylgalactosamine-6-Sulfatase (RHGALNS) in Pediatric Patients Less Than 5 Years of Age With Mucopolysaccharidosis IV A (Morquio A Syndrome, MPS IVA)

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Preliminary results after 26 weeks of treatment from an ongoing study evaluating safety and efficacy of RHGALNS in 15 patients with MPS IVA <5 years of age are reported. The mean (range) age was 3.1(0.8-4.9) years. Standing height/length ($n = 15$) was severely affected in many patients; 7 (46.7%) at <third, 3 (20.0%) at >3rd to <10th, 2 (13.3%) at >25th to <50th, and 3 (20.0%) at $\geq$50th percentiles. The most commonly reported adverse events (AEs) were vomiting in 12 (80.0%), pyrexia in 11 (73.3%), and cough in 8 (53.3%) patients. The majority of AEs were mild to moderate with 1 severe event of tonsillar hypertrophy. No patients discontinued due to an AE. The RHGALNS treatment had a similar safety

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