become available and treatment decisions will have to be take in such cases.

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Screening for MPS VI in a high-incidence area of Northeast Brazil: Report of the first 1,000 newborns tested

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Mucopolysaccharidosis VI (MPS VI) is caused by deficiency of N-acetylgalactosamine 4-sulfatase (ARSB) resulting in storage of dermatan-sulphate in lysosomes and leading to progressive and severe bone dysplasia and to problems in many organs and systems. MPS VI is a very rare condition, which has a relatively high incidence in the county of Monte Santo, in Northeast Brazil (50,000 inhabitants, with 13 MPS VI cases identified thus far). A common mutation (H178L) was identified in all cases. As MPS VI can be treated with ERT and as there are indications that a better outcome may be expected in early treated cases, a newborn screening program for MPS VI was added to the program already in place for PKU, hypothyroidism and hemoglobin disorders, including ARSB assay and detection of the common mutation on DBS. The project started in January, 2011 and a total of 1,000 samples were analyzed through June, 2012. During this period, 55 samples showed ARSB enzyme activity below normal reference values, and 19 of them presented the specific mutation in heterozygosity. With these results we conclude that the methodology developed for MPS VI newborn screening is effective for early detection of the disease in the studied region. The frequency of heterozygotes found for the population of Monte Santo was considered significant, taking into account the disease rarity. The program is in progress, as an increased sample size will allow us to estimate more precisely the frequency of the mutation and the expected number of heterozygotes and homozygotes in the region, which will be important for a population medical genetics approach to the community, including genetic counseling, prenatal, diagnosis and early treatment.

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The incidence of p.R506Q and c.G20210A mutations in South Brazilian patients with Fabry disease and with Gaucher disease

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The incidence of venous thromboembolism (VTE) is ~1 per 1000 annually in the adult general population. A hereditary or acquired cause or even both could be identified in over 80% of the cases. The factor V Leiden (FVL) mutation is the most prevalent inheritable risk factor for VTE. Heterozygous carriers of FVL have an approximately 3-to 5-fold increased risk, whereas the risk in homozygous patients is up to 80-fold higher. Other less common inherited coagulation abnormalities include deficiencies of protein S and C, prothrombin mutation (c.G20210A) and elevated levels of clotting factor VIII.

Objectives: Our aim was to genotype all patients with Gaucher and Fabry diseases followed at Hospital de Clínicas (HCPA), in Porto Alegre, Brazil, regarding the FVL (p.R506Q) and prothrombin mutations.

Methods: We analyzed all patients by Real Time PCR (TaqMan® Genotyping) and confirmed the results by RFLP-PCR (restriction endonucleases: Hind III and MspI).

Results: These 2 mutations were analyzed in 44 patients with Fabry disease and 38 patients with Gaucher disease previously diagnosed by enzyme assay and molecular analysis. The mutation c.G20210A was found in heterozygosis in one Fabry patient (2.2%) and in one Gaucher patient (2.6%). The mutation p.R506Q was found in heterozygosis in four Gaucher patients (10.5%).

Conclusions: The analysis of these mutations in Fabry and Gaucher patients is important since these subjects present a procoagulant state that can lead to transient ischemic attacks, strokes and avascular necrosis of extremities. The heterozygosity of these mutations could be potentially related to the comorbidities presented by these patients.

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The condition of hearing in patients with varying types of mucopolysaccharidosis (MPS)

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The degree of hearing impairment in children with mucopolysaccharidosis (MPS) can vary from grade I to deafness depending on the type and stage of the pathology. Mild and moderate hearing impairment as well as otitis media with effusion-induced hearing impairment often remain undiagnosed due to the changed personality in MPS patients, which is why the hearing of MPS children should be tested no less than twice a year even if there are no complaints. Conventional techniques are employed to assess the condition of hearing in MPS children, i.e., registration otoacoustic emission, impedancometry, registration of evoked acoustic potentials, tonal threshold audiometry or speech audiometry in game form. Ongoing studies of 14 children with MPS (11 boys and 3 girls, aged 2 through 17 years). One child had MPS I, 6 children had type MPS II (Hunter syndrome), 4 children had MPS III (Sanfilippo syndrome), 2 had MPS IV, and 1had MPS VI. Five children had normal intellect, whereas the rest suffered from varied intellectual deficiency. The 13 children had impaired hearing of varying severity from mild dysfunction of the Eustachian tubes to grade III-IV mixed hearing impairment. Five children underwent endoscopic adenotomy. Two children underwent adenotomy and tympanostomy. Three children had hearing aids.

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