

## Abstract Template

Approaching Clusters of Mucopolysaccharidoses in Latin America with Population Medical Genetics Tools

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**Background & Objectives** The Mucopolysaccharidoses (MPS) are rare genetic disorders caused by deficiency of lysosomal enzymes resulting in the accumulation of undegraded glycosaminoglycans (GAGs). The combined incidence of the MPS subtypes in the general population is estimated as 1:25,000 live births. Clusters of these diseases have been identified in areas with high consanguinity rates and/or founder effect associated to endogamy. **Method (s) and Results:** The MPS Brazil Network, associated to the Brazilian Institute of Population Medical Genetics (INAGEMP), identified several MPS clusters in Latin America and investigated them by biochemical and molecular analyses. Three clusters were confirmed in Brazil, of MPS IIIC (state of Paraíba), MPS IVA (state of Paraíba) and MPS VI (state of Bahia). Two clusters were identified in Ecuador: MPS IIIB (state of Manabi) and MPS IVA (state of Pastaza). A cluster of MPS VI was also identified in the Dominican Republic. Other clusters are being investigated in Haiti (MPS VI), Panamá (MPS IVA), and Brazil (MPS IIIB, Minas Gerais state). Haplotype analyses are underway, and results already available for the clusters of MPS IVA and MPS VI in Brazil indicate founder effects with common ancestors. As one example of the benefits of cluster identification, a newborn screening program for MPS VI was implemented in the specific Brazilian region to provide early



identification and treatment. Measures to increase awareness of the community, to provide training to health care personnel, as well as genetic counseling and prenatal diagnosis, could also be offered. **Conclusions (Significance and Impact of the Study):** Examples of MPS clusters were identified in Latin America, and likely, several others are still unreported. The identification and characterization of MPS clusters provides a better understanding about how they were originated and it also enable preventive and management measures to the affected communities.

**Conflict of interest disclosure:** The authors declare no potential conflicts of interest, whether scientific, financial and personal.

**Keywords:** Population medical genetics, mucopolysaccharidoses, clusters, lysosomal storage disorders, inborn errors of metabolism.