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Frequency of mucopolysaccharidoses diseases at the Hospital Infantil de México Federico Gómez

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INTRODUCTION

The mucopolysaccharidoses (MPS) are lysosomal storage diseases due to the deficiency of a given enzyme that normally should undertake the degradation of glycosaminoglycans (GAG). The specific enzymatic deficiencies cause the accumulation of GAG in the lysosome with subsequent organic dysfunction and are abnormally excreted in the urine.[1]The MPS are classified into seven types and are inherited as autosomic recessive diseases with the exception of MPS II that has an X linked recessive pattern of inheritance. The clinical evolution of the MPS is chronic and progressive, with a variable presentation in age and severity; the clinical characteristics may not be evident at birth. Most of the MPS have a multisystem effect: the patients have facial coarseness, organomegaly and multiple dysostosis. The diagnosis is based on the clinical manifestations, radiological and laboratory examinations. The frequency of MPS as a group is about 1:22,500 new born.

As far as we know, there are just a few published reports of Mexican patients suffering from the different types of MPS; a recent presentation from two hospitals in Veracruz in the 2011 Mexican Meeting of Human Genetics reported that they have registered 21 MPS cases in a 7 years period, more than half of them corresponding to MPS II.[2] Another communication reports the analysis of 53 Mexican patients with MPS I.[3] On view of the new therapeutic approaches (enzymatic replacement therapy for example for some types of MPS) we considered with interest to determine the frequency and type of MPS cases that have been attending to our Institution, the Hospital Infantil de México Federico Gomez which was founded 68 years ago and is one of the leading Mexican Pediatric Hospitals.

We had two information sources: the first corresponded to two thesis reviews carried out in 1975 and 2006; the second source was the medical records from 2006 of MPS patients currently attending to our Hospital. The registries included patients from 1943 to 2010, however from 1974 to 1994 there was not possible to retrieve information as there were not records available. Accordingly to the report by Gamboa[4] in 1975 (Registry 1) a review of MPS cases were carried out from 1943 (the founding year of our hospital) to 1974. Fuentes[5] included in her review the patients diagnosed from 1994 to 2005 (Registry 2) and the current medical records included the patients identified between 2006 and 2010 (Registry 3).

The number of patients, gender and MPS type were documented only when there was a confirmed diagnosis of MPS accordingly to the medical and laboratory resources available at the time. In the Registries 1 and 2 the diagnosis was established mainly upon the clinical radiological and biochemical records and in the Registry 3 there were also included (in some cases) the enzymatic activity level, the GAG specific type in urine and the mutation analysis. The information was analyzed by percentages and descriptive statistics analysis (Table 1).

Table 1. MPS patient's gender distribution according to type of MPS.

Registry	Diagnosis form	MPS I		MPS II		MPS III		MPS IV		Total
		Male	Female	Male	Female	Male	Female	Male	Female	
Registry I	Clinical, radiological and urinary GAGs	2	1	1				2	2	8
Registry II	As in Registry I	5	5	1	-	1	5	2	-	19
Registry III	Clinical, radiological, urinary GAG (specific type), enzymatic activity level and in some cases mutation analysis	-	2	9	-	-	-	-	-	11
Total		7	8	11	-	1	5	4	2	38

Care was taken to confirm that the cases were registered only once.

The final number of patients with MPS identified in the three registries was of 38. It was determined that 15 out of 38 cases (40%) corresponded to MPS I; 11 cases corresponded to MPS II (28%), 6 cases to MPS III (16%) and 6 to MPS IV (16%). There were 5 familial cases identified that corresponded to MPS IV (3 cases) and to MPS II (2 cases).

Our results shows that the most frequent MPS in our population was MPS I, followed by MPS II and in third place both MPS III and IV. There were no other MPS types diagnosed; we may assume that in accordance to the low international frequency reported, they are also rare in our population, in particular when considering that in our hospital during 2010 about 140,000 medical consultations were offered. The gender difference among the patients was expected accordingly to the patterns of inheritance, and although there are a higher number of males affected, they correspond to familial cases of MPS II.

In conclusion, this report allowed us to know the frequency profile for MPS of the patients managed at our institution, one of Mexico's reference medical pediatric centers over a period of almost 50 years. The risen incidence for MPS cases during the last decade (as seen also in other recent reports) is probably the result of a wider awareness among the medical community of the MPS diseases, a situation probably related to the interest to offer to the patients for some MPS types the new therapeutic approaches and genetic counseling supported by an accurate enzymatic and molecular genetic diagnosis.

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Recibido el 24 de junio 2011.
Aceptado el 6 de junio 2012.