RareGen Portal

`THE CONCEPT’

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Problems

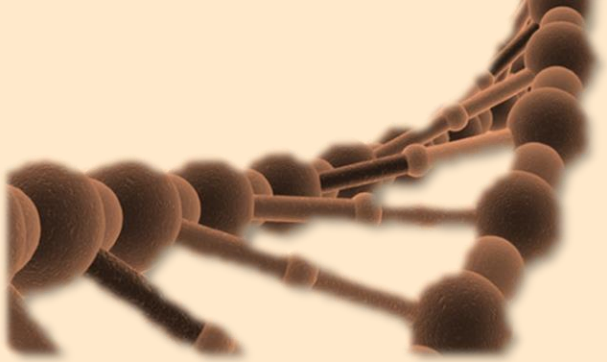
Problem 1

Professionals must get the correct diagnostic for a condition they might have never even listened about.

Problem 2

Diagnosed patients have to undergo a process of acceptance, normally completely alone, with no one that understands what they are going through.

Objectives



Information from healthcare practitioners

Described mutations, symptomatology and possible tests for rare diseases

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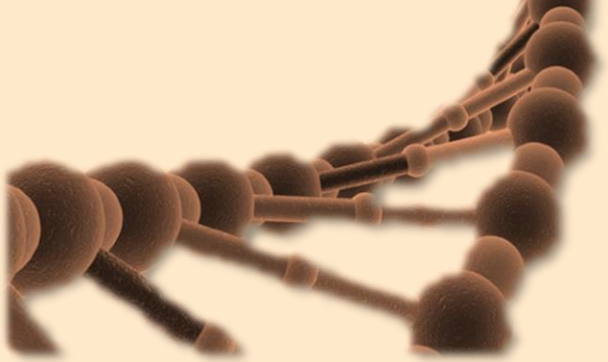

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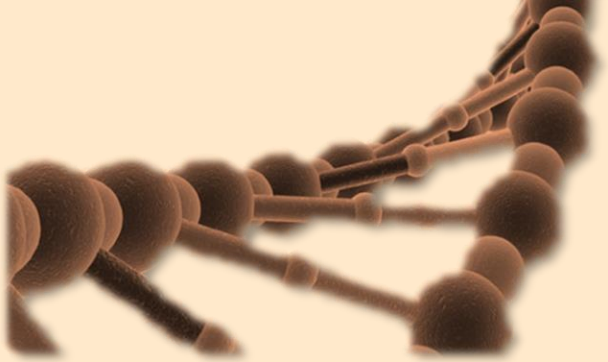

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Patient Associations

Redirection to patient associations classified by country

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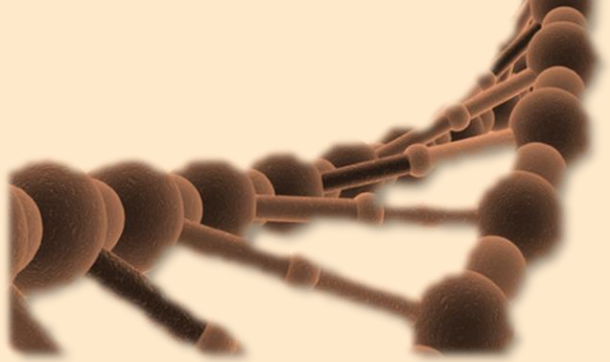

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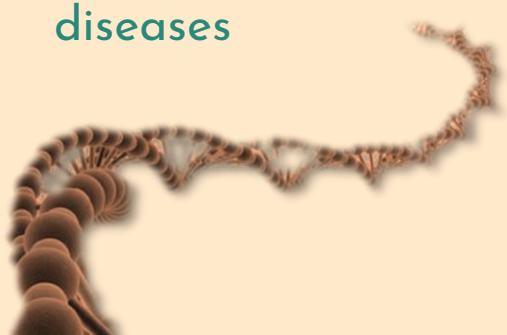
Patient experiences

First-hand accounts from patients



Databases

Lysosome

Mucopolisacaridosis → 

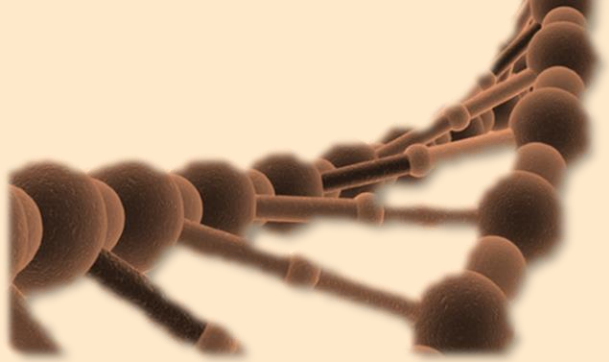
We will use a **mySQL** database created by ourselves with information obtained from

MPS España

Orphanet

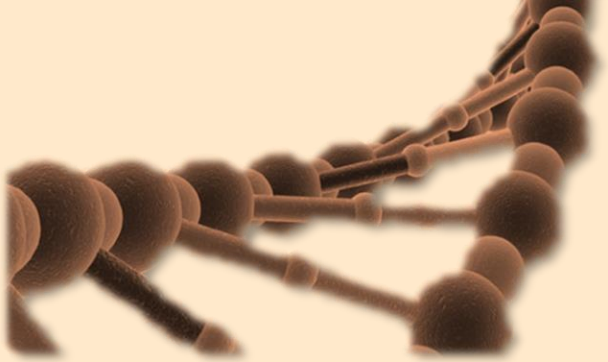
GeneReviews

Input



Search	Type
Hurler syndrome	**Disease** ▲
	Disease
	Gene
	Mutations

Output



Hurler's Syndrome

- Name: Hurler's Syndrome or Mucopolisacaridosis type I
- Gene: *IDUA* (4p16.3)
- Protein: alpha-L-iduronidase enzyme
- Inheritence: Autosomal, recessive
- Prevalence: 1-9/1.000.000
- Symptomatology: There is a delay of the motor and cognitive development, with muskuloskeletal alterations. The age of onset is 3-6 months after birth, however, there is an intermediate form in which the onset is in Adulthood (which presents normal stature and no intellectual deficit has been observed)...
- Diagnostic method: Detection of an increase of heparan and dermatan sulfate in urine, detection of alpha-L-iduronidase enzyme in leukocytes, genetic testing
- Treatment: Enzyme replacement treatment
- ...

THANKS FOR YOUR ATTENTION QUESTIONS?