

# MPS I Survey Results

## Patterns in the Referral, Diagnosis, and Management of Individuals with MPS I

A collaboration between The National MPS  
Society and Genzyme Corporation

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## Survey Objectives

- **To learn from individuals with MPS I and their families about their experiences with diagnosis and management**
- **To discover the challenges and barriers faced on the path to diagnosis**
- **To obtain a basic overview of the experience of MPS I patients and their families regarding the treatment and management of the disease**
- **To use learnings to help design education and outreach programs related to MPS I disease**

## Survey Methodology

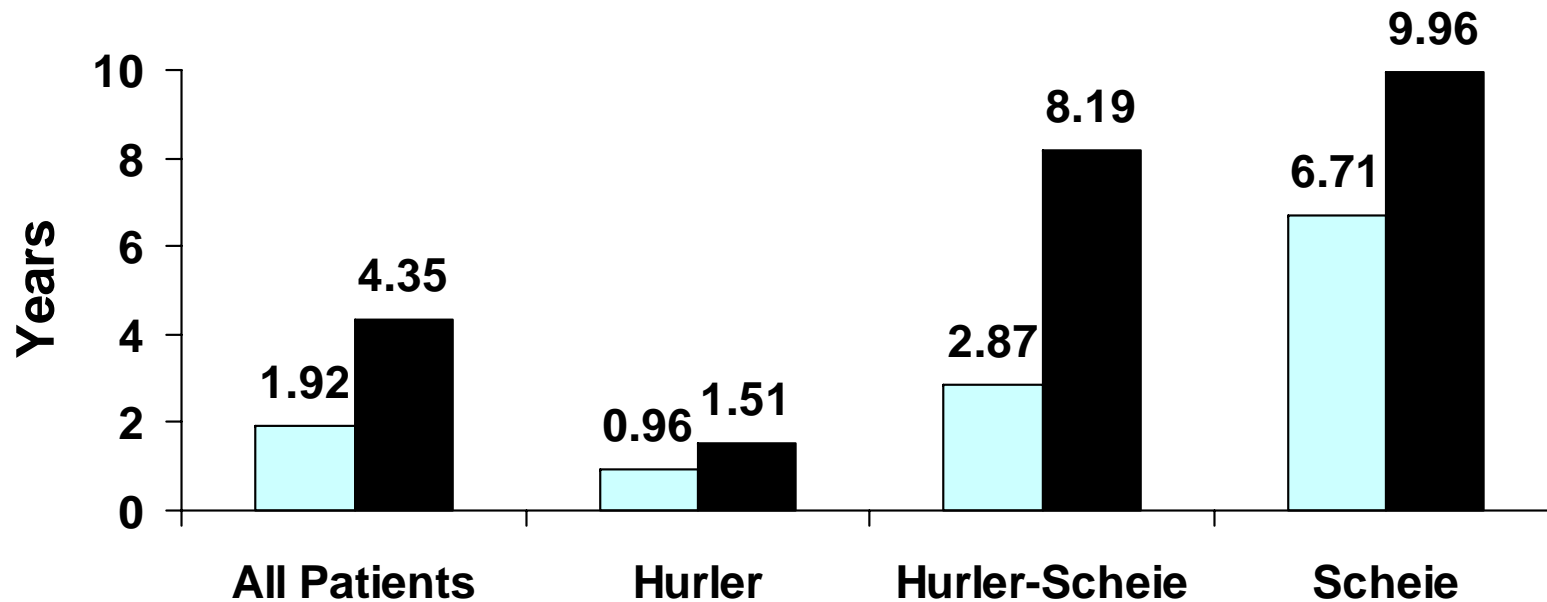
- **8 page questionnaire with 12 broad questions that took approximately 15-30 minutes to complete**
- **Mail survey sent by the National MPS Society to approximately 100 members of National MPS Society and non-member individuals and families in the July-August 2003 timeframe**
- **65 responses received, mostly in the September-October 2003 timeframe**
- **Responses were optional, anonymous, and confidential**
- **Responses were analyzed as a group only**

## Respondent Profile

- **Person completing questionnaire**
  - Parent of individual with MPS I ..... 89%
  - Individual with MPS I ..... 11%
- **MPS I subgroup**
  - Hurler..... 55%
  - Hurler/Scheie..... 29%
  - Scheie..... 16%
- **Gender of individual with MPS I**
  - Male..... 59%
  - Female..... 41%
- **Current age of individual with MPS I**
  - 13 years (on average)

**It was reported that on average it takes 2 ½ years from the first time people visit a physician regarding symptoms until MPS I is suspected. This time lag appeared to be longer for Hurler-Scheie and Scheie patients.**

**Age at first presentation vs Age at first suspicion of MPS I  
(average number of years)**

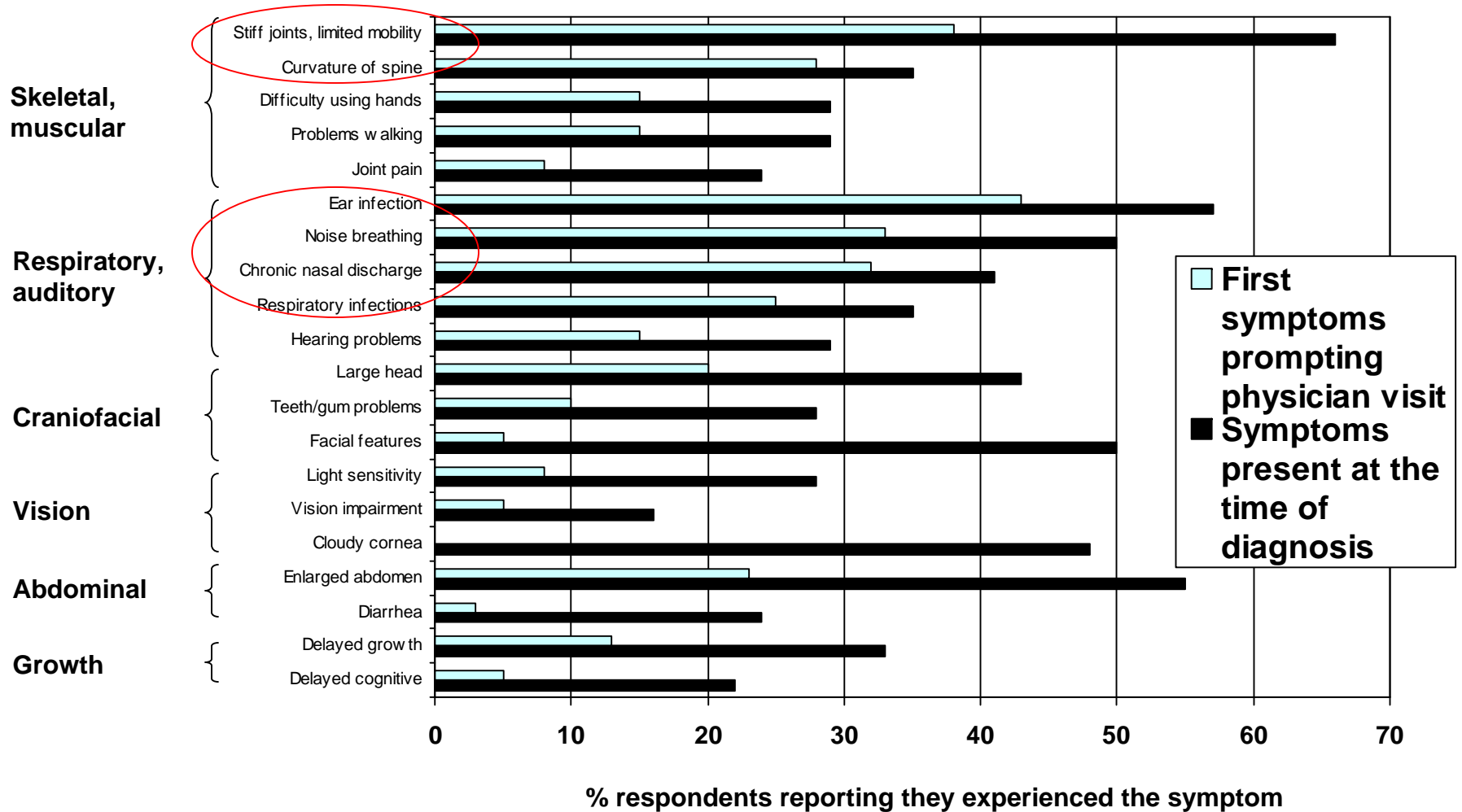


■ Age when first visited physician  
■ Age when a physician first suspected MPS I or another genetic disease

\* List is not comprehensive but represents the collection of physician specialists that in general represented the majority of responses.

+ In some cases, several additional visits were required until diagnosis was made. However, most results fall within the range of 1-8 physicians

**It was reported that some of the most common initial symptoms that individuals with MPS I present with to physicians were the same as those of other common pediatric illnesses (e.g. ear infections, runny noses)**



**It was reported that after the initial visit to their pediatrician or family/ general practitioner (GP), individuals with MPS I may need to visit several other types of specialists before a final diagnosis is made**

**Physicians Visited by MPS I Patients\***

Order of Visits to Physicians <sup>+</sup>	Pediatrician	Family / General Practitioner	Ear, nose and throat (ENT)	Ophthalmologist	Orthopedic surgeon	Pulmonologist	Neurologist	Cardiologist	Geneticist	Metabolic Specialist
	1 <sup>st</sup> physician seen (100% of responses)	62%	21%	5%	2%	2%	2%	2%	2%	2%
2 <sup>nd</sup> physician seen (91% of responses)	19%	5%	19%	3%	13%	7%	3%	7%	15%	--
3 <sup>rd</sup> physician seen (93% of responses)	17%	5%	19%	2%	9%	4%	4%	11%	20%	2%
4 <sup>th</sup> physician seen (93% of responses)	12%	2%	10%	16%	6%	4%	10%	4%	27%	12%
5 <sup>th</sup> physician seen (62% of responses)	11%	--	8%	17%	6%	3%	11%	11%	22%	3%
6 <sup>th</sup> physician seen (100% of responses)	11%	--	7%	14%	17%	4%	14%	11%	25%	4%
7 <sup>th</sup> physician seen (94% of responses)	40%	--	--	7%	7%	7%	13%	--	20%	--
8 <sup>th</sup> physician seen (99% of responses)	33%	--	11%	--	--	--	--	11%	44%	--

\* List is not comprehensive but represents the collection of physician specialists that in general represented the majority of responses.

<sup>+</sup> In some cases, several additional visits were required until diagnosis was made. However, most results fall within the range of 1-8 physicians.

## In the path to diagnosis, it was reported that many physicians either take no action or simply monitor the individuals

- Many physicians simply monitor the symptoms they were referred for, but may not be putting the entire picture together to make a diagnosis of MPS I
- If a referral to geneticist or metabolic specialist is made, then there is a higher likelihood of a correct diagnosis

For each physician seen, please select from the following to indicate the action(s) taken by the physician or you

	Pediatrician	FP/GP	ENT	Ophthalmologist	Orthopedic Surgeon	Pulmonologist	Neurologist	Cardiologist	Geneticist	Metabolic Specialist
No action, taken. Physician reassured patient that nothing was wrong.	23%	21%	--	4%	3%	17%	13%	5%	--	--
Physician monitored or managed the symptoms.	30%	33%	77%	46%	48%	50%	38%	53%	4%	13%
Physician made a referred to somebody else.	18%	29%	6%	8%	17%	17%	6%	11%	4%	13%
Physician suspected a genetic disease and referred to somebody else for confirmation.	16%	8%	--	23%	13%	17%	13%	5%	9%	--
Physician made a diagnosis other than MPS I.	7%	4%	12%	4%	10%	--	6%	21%	6%	--
Physician made a diagnosis of MPS I.	4%	--	--	12%	3%	--	19%	5%	77%	75%
Don't remember what physician did.	1%	--	--	--	--	--	--	--	--	--
I went to another physician for a second opinion	2%	4%	6%	4%	7%	--	6%	--	--	--
	100%	100%	100%	100%	100%	100%	100%	100%	100%	100%

**It was reported that individuals with MPS I were diagnosed as having a variety of conditions on their way to a final diagnosis\*+**

- **Allergies, asthma or reactive airway disease**
- **Colic**
- **Laryngomalaysia**
- **Meningitis**
- **Marfan's syndrome**
- **Maroteaux-Lamy (MPS VI)**
- **Scleroderma**
- **Muscular dystrophy**

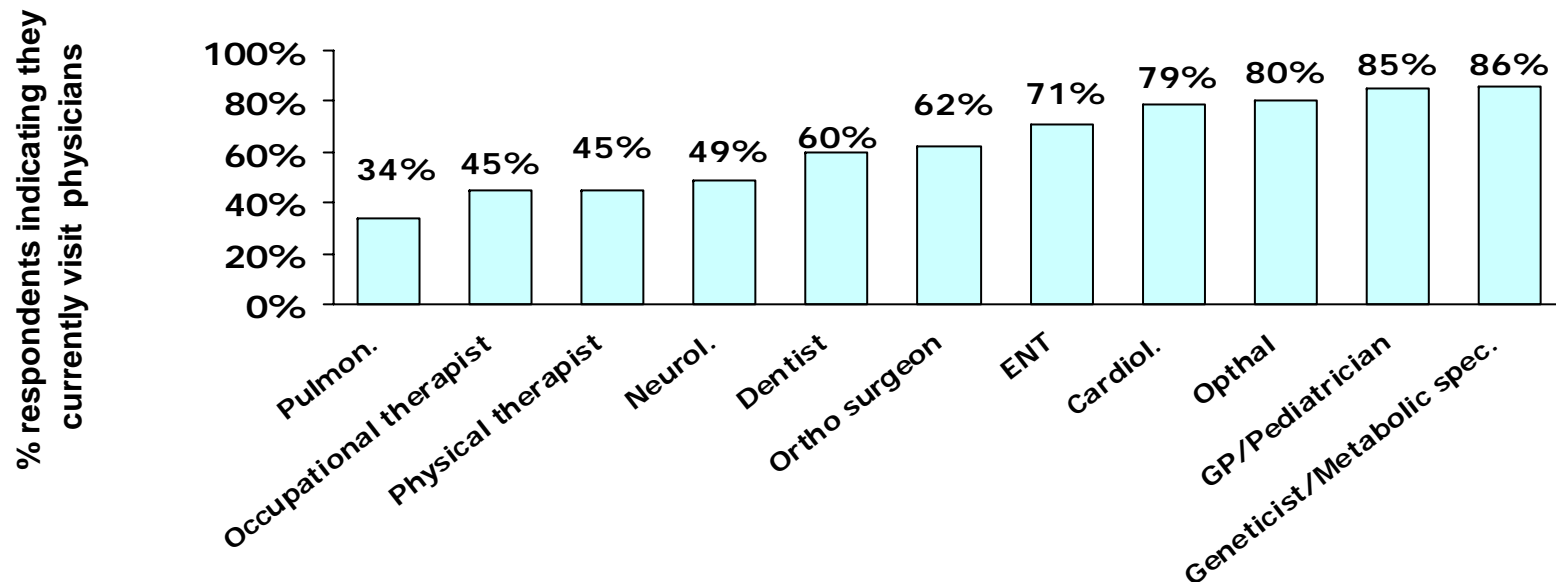
\* List is not comprehensive but represents a sample of responses.

+ Due to the highly variable presentation of the symptoms of MPS I, and their similarity to those of other disorders, these results should not be interpreted as highlighting “misdiagnosis” of patients

## It was reported that on average individuals with MPS I currently see seven different physicians for symptoms associated with MPS I

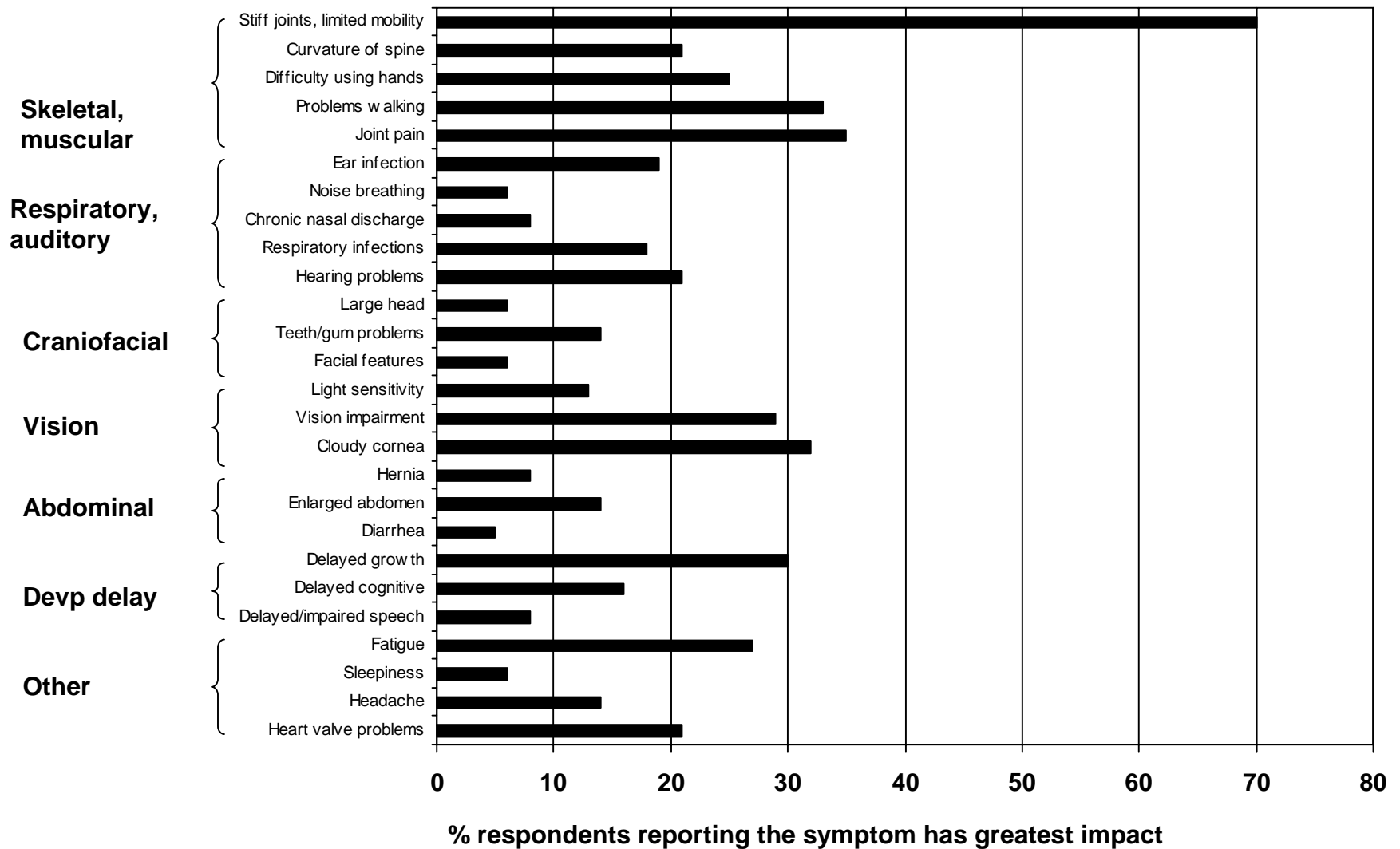
- Variation in total number of physicians involved is quite large:
  - 1-2 physicians seen (21%)
  - 3-6 physicians seen (25%)
  - 7-9 physicians seen (39%)
  - 10+ physicians seen (23%)
- 73% of individuals report having a primary care physician who manages or coordinates their overall medical care (mainly pediatricians and geneticists)

Physicians Currently Visited\*

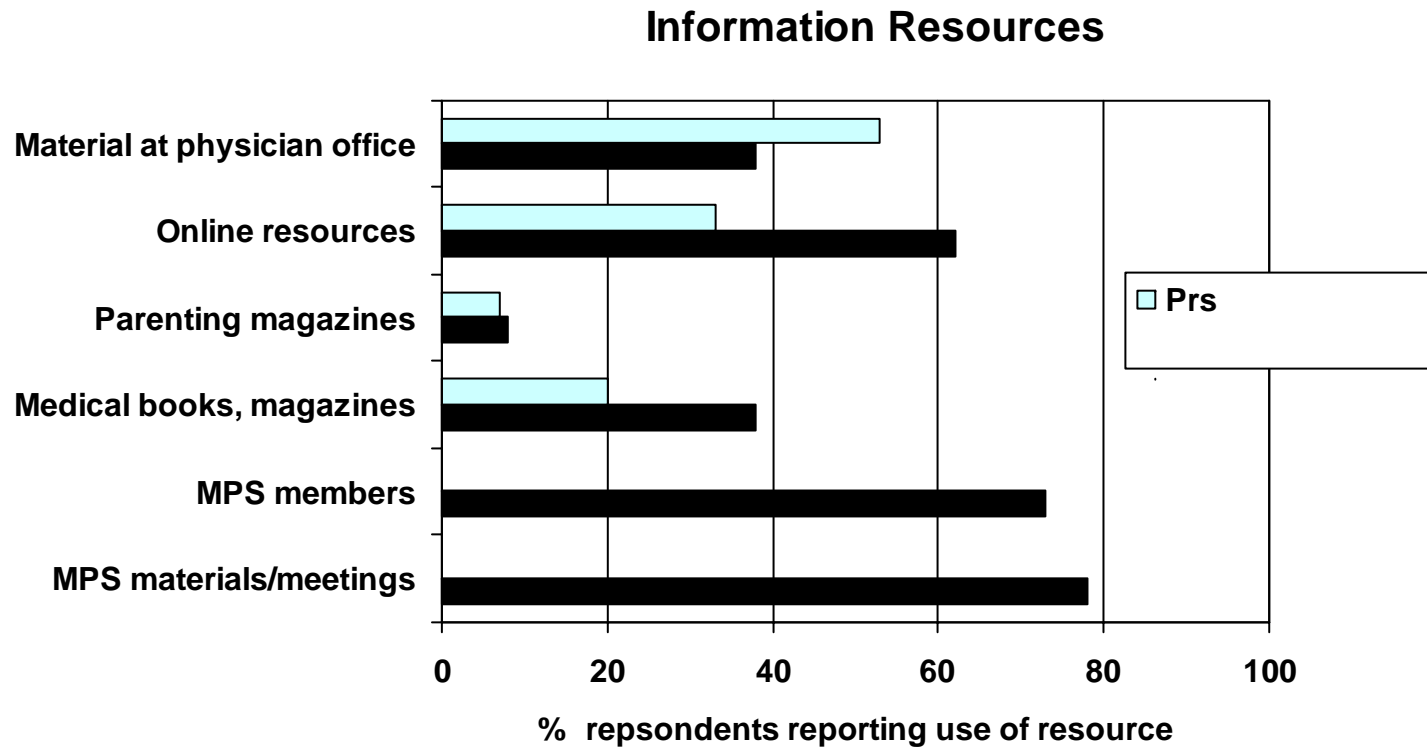


\* List is not comprehensive, only top 11 specialists mentioned. Other physicians specialists include Hematologist/Oncologists, speech therapists, general surgeons, general internal medicine specialists, etc.

**It was reported that by far those symptoms related to joint stiffness, pain, and mobility had the greatest impact on individuals with MPS I, followed by symptoms related to visual and auditory impairment.**

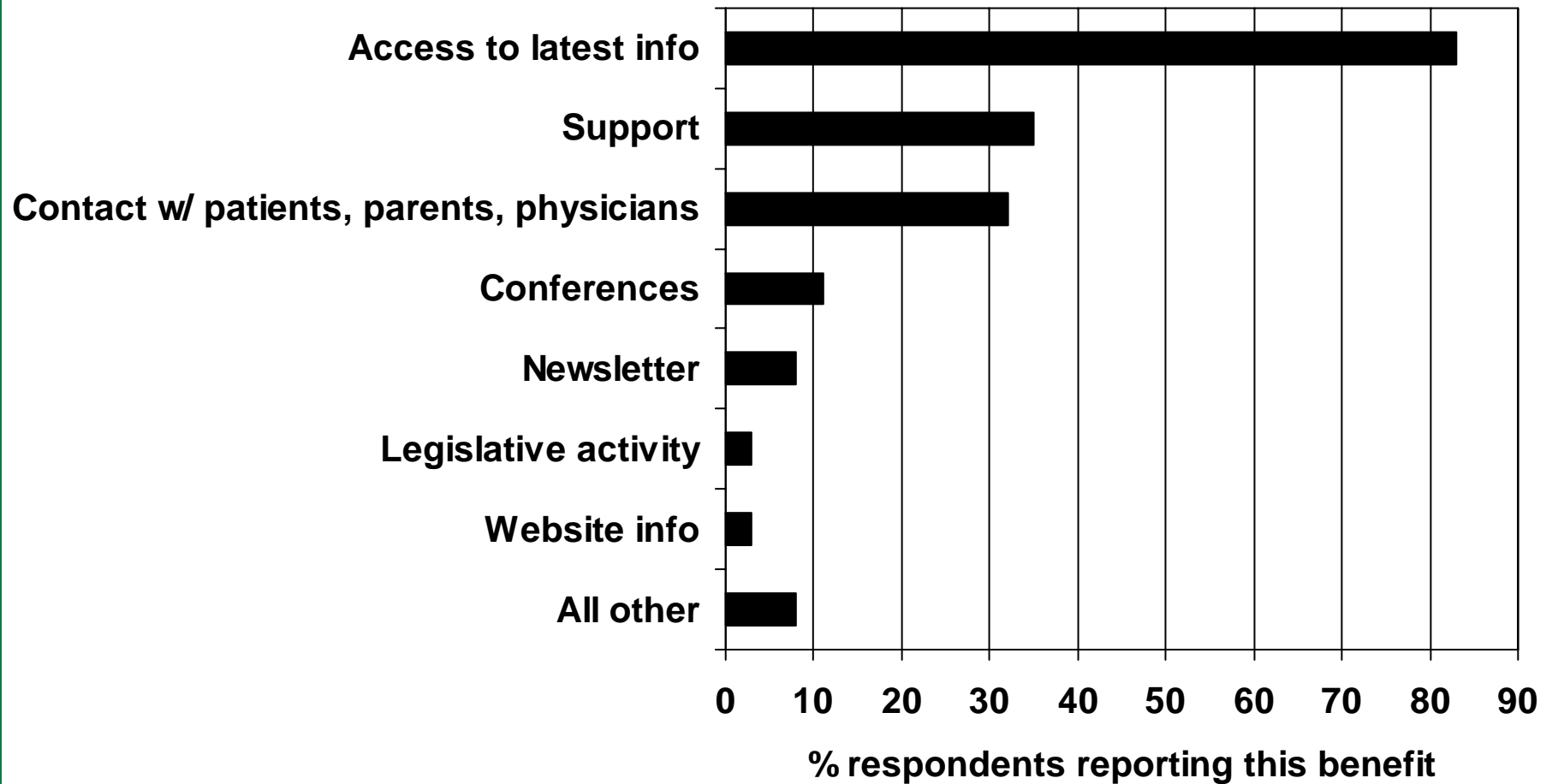


**It was reported that to first learn about MPS I, individuals and their families primarily turn to materials in a physician's office. To learn more after diagnosis most turn to the National MPS Society and other families.**



# Individuals and families report deriving many benefits from National MPS Society membership.

## Main benefit of being a member of the National MPS Society



## Conclusions

- **There is a significant gap between when the early symptoms of MPS I present and when a diagnosis is made**
- **There is a need to raise awareness among physicians about the early symptoms of MPS I**
  - **MPS I is a highly multi-systemic disease, but these symptoms may be difficult to differentiate from those of common illnesses**
  - **Individuals maybe be diagnosed as having many conditions other than MPS I**
  - **Physicians may not be recognizing the cluster of symptoms that characterizes MPS I disease**

## Conclusions

- **There is a need to increase the sense of urgency among pediatricians to refer patients with unusual symptoms to geneticists or metabolic specialists**
  - Many different physicians may be involved in the referral path to eventual diagnosis, but pediatricians and FPs/GPs are usually among the first to be consulted
  - Many physicians may be simply monitoring symptoms rather than making a referral
- **Many different physicians may need to be involved to manage the symptoms of individuals with MPS I**
  - Pediatricians and geneticists are most often the ones that coordinate overall care with a multidisciplinary team

## Conclusions

- **Individuals with MPS I and their families seek information from many different sources prior to and after diagnosis**
  - **The National MPS Society is the preferred source for information for those that become members, and members derive many benefits from their involvement with the Society**