Quality of life assessment in mucopolysaccharidosis patients treated with enzyme replacement therapy

Mariem AJILI(1), Mouna ZRIBI(1), Hela BOUDABBOUS(1), Sana BEN MRAD(1), Zaki HIDOURI(1), Mohamed Slim ABDELMOULA(1), Amel BEN CHEHIDA(1) and Neji TEBIB(1)

(1) La Rabta Hospital Tunis, Tunisia
**Introduction**

Mucopolysaccharidoses (MPS) are a group of orphan lysosomal diseases.

**Clinical manifestations** include:
- Variably coarse features
- Mental impairment
- Enlarged liver and spleen

Only few studies have focused on physical and psychosocial impacts of these diseases.
**Aim**

To assess the **health related quality of life** (HRQOL) in **children with MPS** treated with **enzyme replacement therapy** in pediatric department of La Rabta Hospital.

**Methods**

MPS < 18 years old  
Treated with enzyme replacement therapy

Pediatric Quality Of Life inventory  
4.0 Generic Core Scale (PedsQL)

- Parent-report (all patients)  
- Self report (when applicable)  
- **Physical functionning**  
- **Psychosocial functioning** (emotional/social/school)
**Results**

- **Age:** 4 to 15 years.

- **Mean age at the start of enzyme replacement therapy:** 9 years and 8 months.

- **Average follow-up:** 2 years and 6 months [2 months - 4 years].

- **Parent’s report PedsQL:** mean total PedsQL score = 66.52/100:
  - Worst score: physical domain (53.51).
  - Highest score: psychosocial domain (72.81/100).

- Scholar functioning score was low in 4 patients (48.75).

- **Self report PedsQL (2 patients):**
  - Physical domain: 76.86/100
  - Psychosocial domain: 85.83/100
Discussion and conclusion

HRQOL of MPS patients in our series was poor (PedsQL < 78.6/100) which is in accordance with literature.

- MPS is a disabling disease and significantly impacts the HRQOL of patients and their parents.
- The enzyme replacement therapy was introduced in order to improve the HRQOL of patients.
- HRQOL depend on socio-economic conditions and the delay of the initiation of enzyme replacement therapy.