

MPS Diseases

Facts and Information

MPS diseases are rare genetic diseases that primarily affect children. They cause progressive damage to cells in the body, resulting in severe disability and early death. There are few treatments and no cures. But there is hope.

WHAT ARE MPS DISEASES?

- There are many types of Mucopolysaccharidoses (MPS) and related diseases, each defined by a specific enzyme deficiency, with a range of severity and impacts within each disease.
- People with MPS diseases produce insufficient amounts of certain enzymes that break down materials in body cells.
- This leads to build-up of excess materials in cells, causing damage that increases over time as the materials accumulate, resulting in profound range of physical and cognitive impacts, including intellectual disabilities, heart disease, vision loss, speech and hearing impairment, short stature, stiff joints, and pain, among others.

MPS diseases result in profound and expansive health problems, such as:

- Intellectual disabilities
- Heart disease
- Vision loss
- Speech impairment
- Hearing impairment
- Short stature
- Chronic respiratory problems
- Stiff joints
- Loss of mobility
- Hyperactivity
- Pain
- Sleeplessness
- Shortened life span

PROGRESSION OF MPS DISEASES

- MPS diseases are devastating for children and families both because of the lack of effective treatments and because they are progressive in nature. Babies with MPS diseases are often born looking perfectly healthy. It is only later, as cell damage becomes worse, that parents receive this heartbreaking diagnosis.
- MPS diseases end up affecting nearly every part of the body, damaging the heart, bones, joints, respiratory system and central nervous system.
- All MPS diseases are terminal. Although there are adults living with MPS diseases most children do not survive beyond their teenage years.
- MPS is always changing and progressing. This means that families know that their child's symptoms will continue to worsen.

IMPACT OF MPS DISEASES ON FAMILIES

- Caring for children with MPS is a full-time job.
- Parents must coordinate care with dozens of medical experts.
- Children with MPS need specialized medical equipment, like oxygen pumps, gastrointestinal feeding equipment, therapeutic beds, braces, wheelchairs, and other specialty medical items.
- For these reasons, the costs to care for children with MPS are astronomical.
- The emotional and social impacts are also immense. Having a child with MPS disease affects nearly every aspect of a family's daily life.

Medical professionals required for care of child with MPS include:

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|-----------------|---------------------------|
| ✓ Pediatricians | ✓ Physical therapists |
| ✓ Neurologists | ✓ Speech therapists |
| ✓ Geneticists | ✓ Occupational therapists |
| ✓ ENTs | ✓ Home nursing aides |
| ✓ Orthopedists | |
| ✓ Cardiologists | |
| ✓ Audiologists | |
| ✓ Dentists | |
| ✓ Oral surgeons | |

TREATMENT OPTIONS FOR MPS DISEASES

- There are **no cures** for any of the MPS diseases.
- There are very few treatments, and some MPS diseases have no treatment options at all.
- The treatments that do exist are expensive and difficult for patients and families because they are invasive and require significant time in the hospital.

WHAT THE FUTURE HOLDS FOR MPS DISEASE TREATMENT

- Researchers who study MPS diseases believe that continued and well-funded research can unlock new treatments, and one day, cures for MPS diseases.
- Acceleration of approval of ultra-rare disease therapies holds promise for new investment from biotechnology companies in MPS diseases and other ultra-rare disorders.
- Growing awareness of MPS diseases can attract research talent, funding and public interest.
- Families of loved ones with MPS diseases are hopeful for this future, yet aware of the ticking clock as their child's disease progresses.