Factsheet: MPS (mucopolysaccharidosis)

About MPS

- MPS stands for mucopolysaccharidosis and is a group of rare genetic disorders.[1]
- People with MPS are missing a specific enzyme, which means their bodies cannot breakdown substances properly.[1]
- This leads to an accumulation of these substances in the body's cells, causing progressive damage – affecting appearance, development and the function of various organs of the body.[1]
- There are 14 types of MPS, each with varying symptoms and signs. More information is found below.[2]

About International MPS Day

- International MPS Day is held on 15 May each year, and aims to raise awareness of those living with this rare genetic disorder.[3]
- This International MPS Day, The Australian MPS Society has launched a new campaign - called ***'Rare Beauty: Faces of MPS' -*** to raise awareness and support for Australians living with MPS, their families, friends and carers.
- The new campaign involves a photographic series, which captures images of 10 Australians living with MPS and mucolipodosis (ML) and their families, in the hope of generating greater understanding and visibility of the impact of living with MPS.

About the Australian MPS Society

- The Australian MPS Society (MPS Society) is a non-profit organisation formed by parents, relatives and friends of those living with a range of rare genetic disorders known collectively as the mucopolysaccharide (or MPS) diseases.
- The MPS Society supports individuals affected with MPS / ML, parents of affected children, other relatives and friends. The organisation is governed by a committed Board of Directors elected by its membership, and has strong relationships with medical experts who provide advice on scientific and policy matters.

- The MPS Society, with the support of industry partners, has recently launched a new look website (https://www.mpssociety.org.au/) and branding to encourage more members and supporters of the Australian MPS Community to re-engage with the organisation, its content and support services – with a view to building more active advocates. The new website is more user friendly, provides more information and support and will now allow families and supporters to renew their memberships online.

About MPS Types

About MPS I

MPS I is an inherited disorder that encompasses a wide spectrum of severity. In some people the brain may be affected in combination with physical symptoms; others may develop physical symptoms with no brain involvement. Physical symptoms may include eye and hearing problems, bone and joint malformation, and heart and breathing difficulties.

Clinically, MPS I is divided into three categories: Hurler syndrome, Hurler-Scheie [pronounced *shay]* syndrome, and Scheie syndrome, so named after the doctors who originally described the condition. Hurler syndrome is generally regarded as the most severe and the brain is affected; Scheie syndrome is the most 'mild' [or attenuated]. Many people fall between the two and are referred to as Hurler-Scheie.

MPS I is inherited in what is known as an autosomal recessive manner. In this form of inheritance both parents must carry a copy of the same defective gene and each pass that defective gene to their child. In the case of MPS I, the defect relates specifically to the faulty production of alpha-L-iduronidase enzyme.

About MPS II

MPS II is an inherited disorder that encompasses a wide spectrum of severity. In some people the brain may be affected in combination with physical symptoms; others may develop physical symptoms with no brain involvement. Physical symptoms may include hearing problems, bone and joint malformation, and heart and breathing difficulties.

MPS II is also known as Hunter syndrome, so named after the doctor who first described the condition.

MPS II is inherited in what is called an X-linked recessive manner. In MPS II the gene that codes for deficient production of 2-sulphatase enzyme is located on the 'X' chromosome.

About MPS III

MPS III is an inherited disorder that encompasses a wide spectrum of severity. The brain is the primary site of disease and its function declines with time; physical symptoms may also develop with time, and may include hearing and breathing difficulties.

There are four types of MPS III, known as types A, B, C and D. Recently, type E was described in a mouse but is yet to be reported in humans. MPS III is also referred to Sanfilippo syndrome, so named after the doctor who first described the condition.

All four known forms of MPS III are inherited in what is known as an autosomal recessive manner. In this form of inheritance both parents must carry a copy of the defective gene and each pass that defective gene to their child.

About MPS IV

There are two main forms of Morquio syndrome, MPS IVA and MPS IVB. The usual form of Morquio syndrome is known as MPS IVA to distinguish it from the much rarer B form which is caused by the lack of a different enzyme. Individuals with the B form have similar problems but tend to be more mildly affected.

MPS IVA individuals are missing the enzyme called galactose 6-sulphatase and MPS IVB individuals are missing the enzyme called beta-galactosidase.

About MPS VI

MPS VI is an inherited disorder that encompasses a wide spectrum of severity. Physical symptoms may include bone and joint malformation, hearing and vision problems, and heart and breathing difficulties.

MPS VI is also known as Maroteaux-Lamy syndrome, so named after the two doctors who first described the condition.

Because MPS VI is inherited it is important to seek genetic counselling as there may be implications for other children in the family, future pregnancies and extended family members.

About ML II & ML III

I-Cell disorder and Pseudo-Hurler Poldystrophy are forms of a condition known as mucolipidoses. They are also known as ML II and ML III. Children with ML II/III are missing an enzyme called phosphotransferase.

ML-III was described by Dr. Maroteaux and Dr. Lamy from France. They called it Pseudo-Hurler Polydystrophy as it resembled a mild form of Hurler Syndrome, one of the mucopolysaccharide disorders. "Polydystrophy" means that many organs are abnormal.

References

[1] MPS Society, (2018). *About the MPS and Related Diseases*. [online] Available at: http://www.mpssociety.org.au/about-the-mps-and-related-diseases/
[2] MPS Society, (2018). *Table of Diseases*. [online] Available at: http://www.mpssociety.org.au/about-the-mps-and-related-diseases/table-of-diseases/
[3] MPS Society UK, (2018). *MPS Awareness Day*. [online] Available at: http://www.mpssociety.org.uk/get-involved/mps-awareness-day/#