

Living as an adult with MPS

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Susanne Gerit Kircher

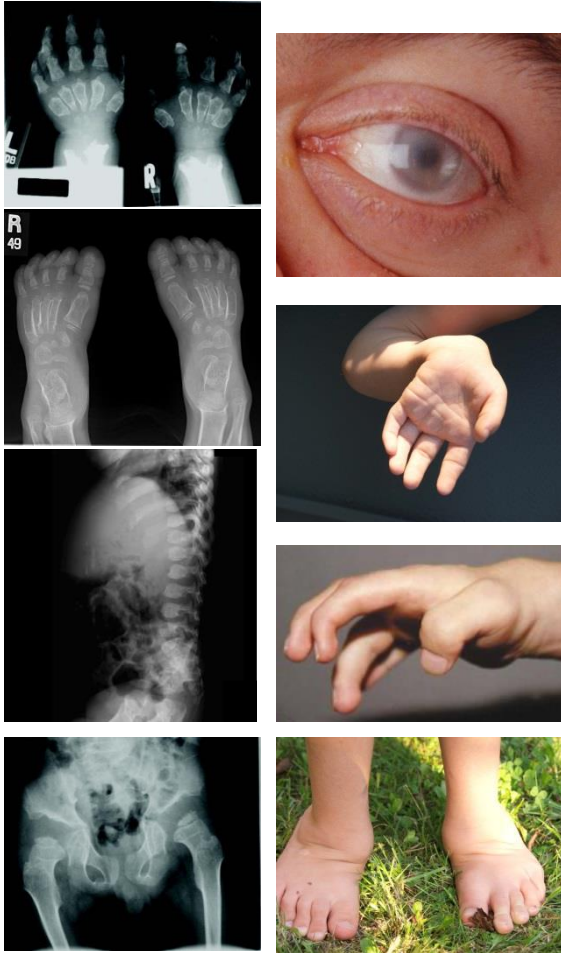
Center of Pathobiochemistry and Genetics,, Medical University of Vienna, Austria

Overview

- ▶ MPS in adults – medical issues
- ▶ Life behind the medicine – childhood, education, professional training and work, family and children
- ▶ Note: The following photos have been available with the kind permission of those with MPS themselves and/or their relatives or of the Austrian Society for MPS. It is expressly requested that no photos of those with MPS be reproduced or circulated.



Characteristics of Mucopolysaccharidoses



- ▶ MPS is a group of **rare genetic diseases** in the extracellular matrix of the connective tissue
- ▶ Connective tissue is represented all over the body – therefore MPS are **multisystemic disorders**
- ▶ Clinical spectrum: from very severe forms with early death to very mild - better: **attenuated - forms** with **survival until late adulthood** (in all types of MPS)
- ▶ MPS cause **increasing organ dysfunctions** with advancing age

Dysostosis multiplex



Mucopolysaccharidoses - types

MPS I H, S	M. Hurler M. Scheie	α-Iduronidase	4p16.3
MPS II	M.Hunter	Iduronat-Sulfatase	Xq28
MPS IIIA	M.Sanfilippo A	Sulfamidase	17q25.3
MPS IIIB	M.Sanfilippo B	N-Acetyl-α-Glucosaminidase	17q21
MPS IIIC	M.Sanfilippo C	N-Acetyltransferase	8p11.1
MPS IIID	M.Sanfilippo D	N-Acetylglucosamine- -6-Sulfatase	12q14
MPS IVA	M.Morquio A	N-Acetylgalactosamine -6-Sulfatase	16q24.3
MPS IVB	M.Morquio B	β-Galactosidase	3p21.33
MPS VI	M.Maroteaux-Lamy	Arylsulfatase B	5q13
MPS VII	M.Sly	β-Glucuronidase	7q21.11
MPS IX	Hyaluronidasedef.	Hyaluronidase	3p21.3



Austrian experiences with the generation 40+

Why don't we know much about these patients?

- After the pediatric care, patients are “lost” to various medical specialists (cardiologists, orthopaedics...)
- Many of those with attenuated forms of MPS do not want to be in contact with the severely affected

Why do we know these persons in Austria?

- (Adult) Meetings of the MPS Society, by chance...



CASE REPORT

Morquio disease in a patient diagnosed as having Perthes disease for 38 years

S.G. Fang-Kircher¹, A. Böck², W. Fertschak³, W. Schwägerl³ and E. Paschke⁴

¹Institute of Medical Chemistry, University of Vienna, Vienna; ²Department of Pediatrics, ³Orthopedic Department, Pulmonological Center of Vienna; ⁴Department of Pediatrics, University of Graz, Austria

*Correspondence: Institute of Medical Chemistry, University of Vienna, Waehringerstrasse 10, A-1090 Vienna, Austria

- ▶ Born: 1939
- ▶ Age of 12: first hip operation due to “bilateral Perthes’ disease”
- ▶ 1992/1993: admitted to hospital for the operation of the second hip (hip- total endoprosthesis)
- ▶ had already hip operation of the other side before
- ▶ Routine x-ray of the chest – dysostosis multiplex
- ▶ Diagnosis: **MPS IVA** (53 years old)
- ▶ Profession: taxi driver

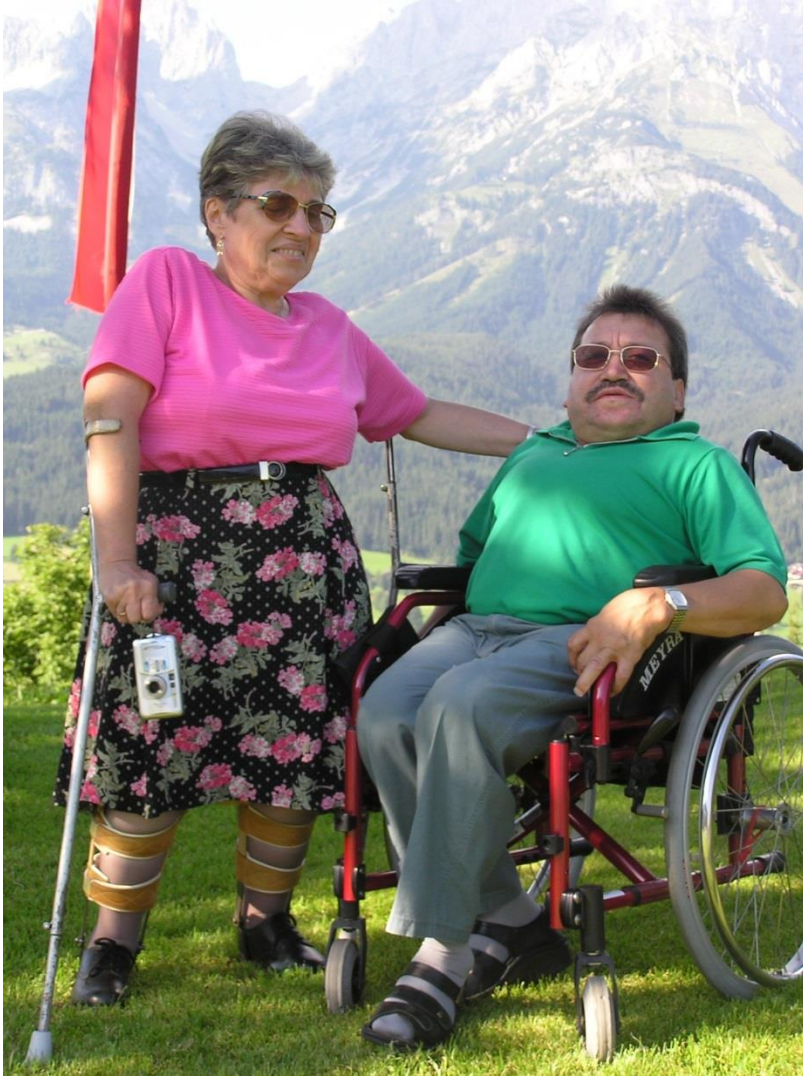


Foto: Dr.Kollar



Foto private

- ▶ Born: 1948
- ▶ 155cm
- ▶ Diagnosis postmortem (by his sister's diagnosis)
- ▶ **MPS VI**
- ▶ Restricted: vision
- ▶ Operations: none
- ▶ Disability: 0%
- ▶ Fulltime job
- ▶ Single
- ▶ No children
- ▶ Died: 1990
(altitude sickness in Peru – lung edema)



- ▶ Born: 1953
- ▶ 140cm, 50-53 kg
- ▶ Suspected: 4 years old
- ▶ Diagnosed: 26 years old
- ▶ **MPS IVB**
- ▶ Restricted: mobility, vision, hearing
- ▶ Operations: hips
- ▶ Disability: 90%
- ▶ Married
- ▶ Died: 2006 (accident)

Foto



Foto private

- ▶ Born: 1954
- ▶ 147cm, 41kg
- ▶ Suspected: at birth (same symptoms as one brother)
- ▶ Diagnosed: 2014 (60 years old)
- ▶ **MPS VI**
- ▶ Restricted: vision, hearing, fine motor skills
- ▶ Operations: heart valves, hands, hips, eyes, cervical spine
- ▶ Disability: 0%
- ▶ retired from a full time job after 41 years
- ▶ Married since 1981
- ▶ No children



- ▶ Born: 1968
- ▶ 147cm, 67kg
- ▶ Suspected: 4-5 years old
- ▶ Diagnosed: 21 years old
- ▶ **MPS IVA**
- ▶ Restricted: vision, mobility
- ▶ Operations: hips, knee, breast
- ▶ Disability: 80%
- ▶ 30 hours / week - job
- ▶ Single
- ▶ No children

Foto





Foto private

- ▶ Born: 1969
- ▶ 150cm, 85kg
- ▶ Suspected: at 1½ years
- ▶ Diagnosed: 2003/2014
- ▶ **MPS IVA**
- ▶ Restricted: mobility, hearing, finger movement
- ▶ Operations: eyes
- ▶ Disability: 100% (since 1997)
- ▶ Fulltime job - mostly via home computer
- ▶ Married since 2006
- ▶ 3 children



- ▶ Born: 1972
- ▶ 142cm, 42.5kg
- ▶ Suspected: 8-9 years old
- ▶ Diagnosed: 19 years old
- ▶ **MPS IVB**
- ▶ Restricted: vision, walking
- ▶ Operations: legs, hips, shoulders
- ▶ Disability 100 %
- ▶ Early retirement (unable to work in the trained profession)
- ▶ Single
- ▶ No children



Foto private

- ▶ Born: 1975
- ▶ 147cm, 41kg
- ▶ Suspected: 3 years
- ▶ Diagnosed: 13 years
- ▶ **MPS IVA**
- ▶ Restricted: vision
- ▶ Operations: hips
- ▶ Disability: 50%
- ▶ Half-time job
- ▶ Second diagnosis since 2 years: multiple sclerosis
- ▶ Married since 2003
- ▶ 2 children



- ▶ Born: 1977
- ▶ 149cm, 49kg
- ▶ Suspected: at birth (same symptoms as sister)
- ▶ Diagnosed: 9 years old
- ▶ **MPS IVA**
- ▶ Restricted: walking, finger movement
- ▶ Operations: hips, knee, elbow
- ▶ Disability: 80%
- ▶ working in an office
- ▶ Married since 2005
- ▶ No children

Foto

First childhood memories

- All symptoms were attributed to „falling from a wall“ on farmers ground
- back brace
- Plaster jacket every night from the age of 3 years, never-ending gymnastics
- Support braces and gymnastics starting at the age of primary school, recurrent middle-ear infections
- 1st operation at the age of 3 years
- Noticed to be „different“, when able to read started to look in magazines and periodicals trying to find someone similar



Education

- ▶ Primary school (1 x 8 years)
- ▶ Secondary school
- ▶ Domestic science college graduate (2x)
- ▶ Apprenticeship (2x, as a dressmaker, shoemaker)
- ▶ Three years of business school, 3-year training to become an industrial manager
- ▶ Grammar school (3x, graduated with Matura)
- ▶ High school for technology (electrical engineering)
- ▶ University (physics, law)



Job title, years of service

- Office, cashier (31 years)
- Dressmaker (early retirement at 30 years of age)
- Shoemaker (23 years)
- Industrial manager (22 years)
- Taxi driver (22 years)
- Head of a department in a governmental office (41 years)
- Electrical engineer / Doctor of Physics, working at University during study, after finishing University assistant, switched to private sector - total more than 20 years
- Doctor of Law, University assistant, deputy head of department at a University institute (18 years)



Voluntary work

- Local community, voluntary fire brigade, church cafe
- „Young Farmers“-group
- Supporting war victims and disabled association
- St. Vincent-Association
- Christian association
- Austrian Student Union
- Representative in the University faculty



What was particularly painful during childhood?

- No one spoke about “being different”, many questions, no answers
- Not able to go skiing or ice-skating with the class-mates
- Weaker than others in sport, quickly exhausted
- Not possible to join the bicycle-tour at the end of the schoolyear
- Numerous operations with stays of up to 4 months in hospital
- When walking with a cane during carneval, was told to „be disabled and not welcome“ by adults
- Was supposed to live for “only 7 years” (heard the medical doctor speaking with the mother because the door was not closed)



Looking back, what is / was painful overall? (1)

- I always have been living independently, now I am occupied so many times a week with my body, all the pains...
- It is not longer possible to go skiing and making sports after hip operation
- My dream of working as a chauffeur or bus driver was not possible to fulfill
- My dream was to learn goldsmith, but the apprenticeship place was too far from home and it was not possible to go there with public traffic every day



Looking back, what is / was painful overall? (2)

- I always wanted to look like my taller healthy brother
- When starting with a new work, every employer expected only a few months of employment, they did not believe in me and my work
- I wanted to have children
- I would be happy to have a child to remember my husband (widow)



What are you proud of?

- Of my wife and children, with whom I can do everything
- Having always been independent
- Having been employed already for 31 years, thus surpassing my personal goal of 25 years
- Having a family and built a house, having my life under control
- Driving my whole life without an accident, once even went to Greece
- Having overcome my shyness and able to talk about my body and my illness
- Having had a profession, a family and having built a house



„Advice“ to parents of those with MPS

- ▶ Allow your child to have a normal childhood
- ▶ do not overprotect it
- ▶ Children should try what they think they can do
- ▶ Always think: first is the child, second is MPS



Advice to the younger people with MPS

- ▶ Try to retain the maximum flexibility: do gymnastics, gymnastics, gymnastics!
- ▶ Believe in yourself, do not say something is impossible
- ▶ Be satisfied with yourself in the face of those more severely affected
- ▶ Accept the situation as it is, make the best of it
- ▶ Don't let it get you down
- ▶ You should not restrict yourself, deny yourself anything, the restrictions come from the others!



Summary: Impressions and thoughts of adult persons with MPS



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schaffen

Desires? Goals? Dreams? Life plans?

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Desires? Goals? Dreams? Life plans?





Institut für Medizinische Chemie
Zentrum für Physiologie und Pathophysiologie

**Institute of Medical Genetics
and Institute of Medical
Chemistry**

**Center of Pathobiochemistry and
Genetics**

Medical University of Vienna



susanne.kircher@meduniwien.ac.at

Tel: +43-(0)1-40160-56512 und -38077, Fax: +43-(0)1-40160-956512

Austrian Society for Mucopolysaccharidoses and Related Diseases



www.mps-austria.at