Adult patients with MPS

- an overview -

Patrício Aguiar

Medicine I Department – Centro Hospitalar Lisboa Norte, EPE

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Disclosures

No conflict of interests related to this lecture.

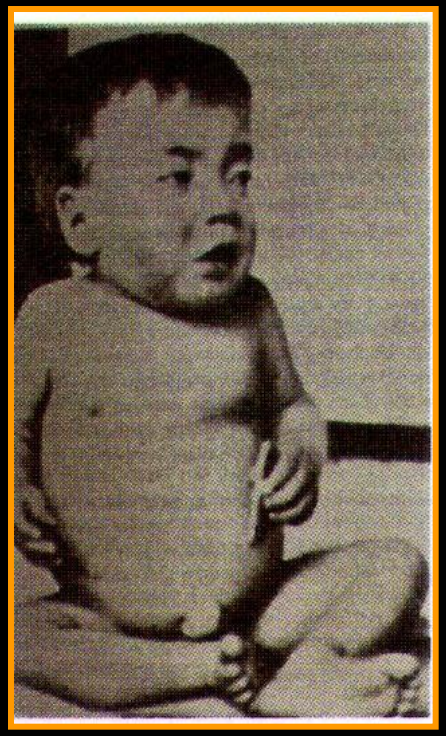
I've received research, congress and travel or advisory grants from Shire, Genzyme and Biomarin.

Mucopolysaccharidosis

Lysosomal storage disorders

inherited error of glycosaminoglycans metabolism

1900



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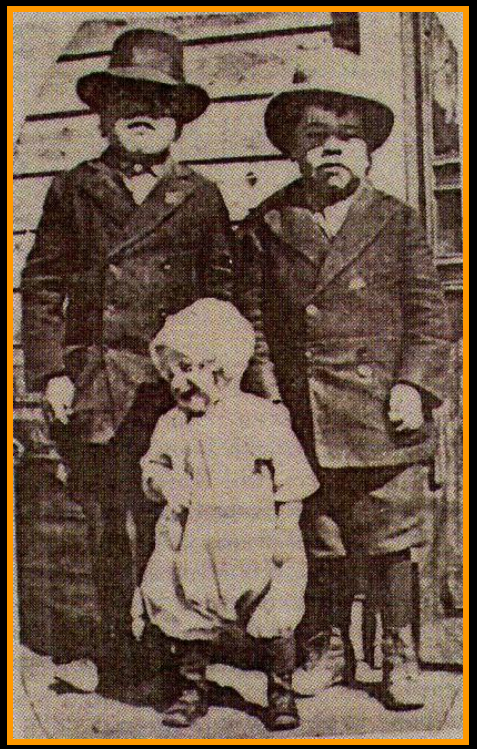

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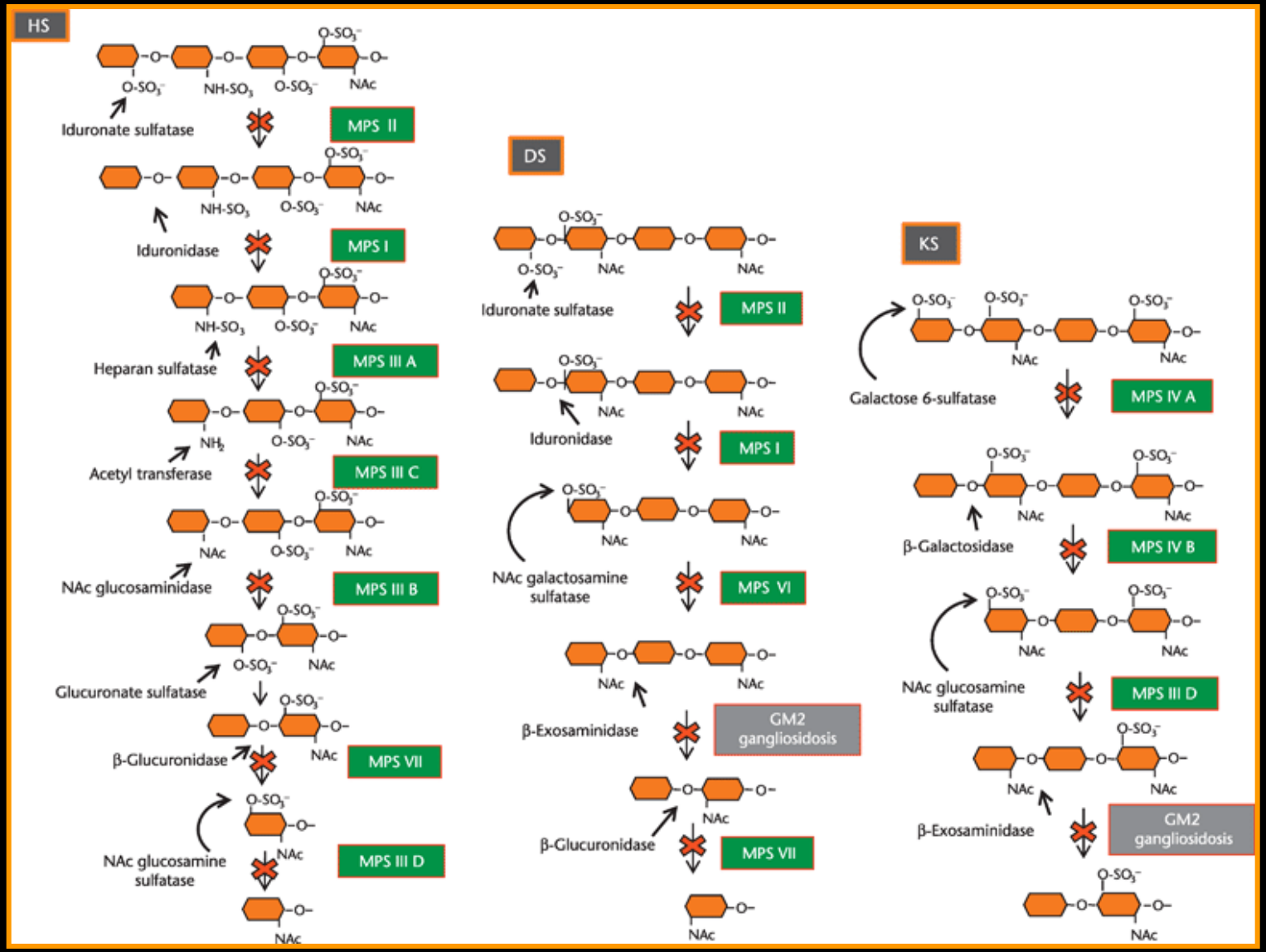
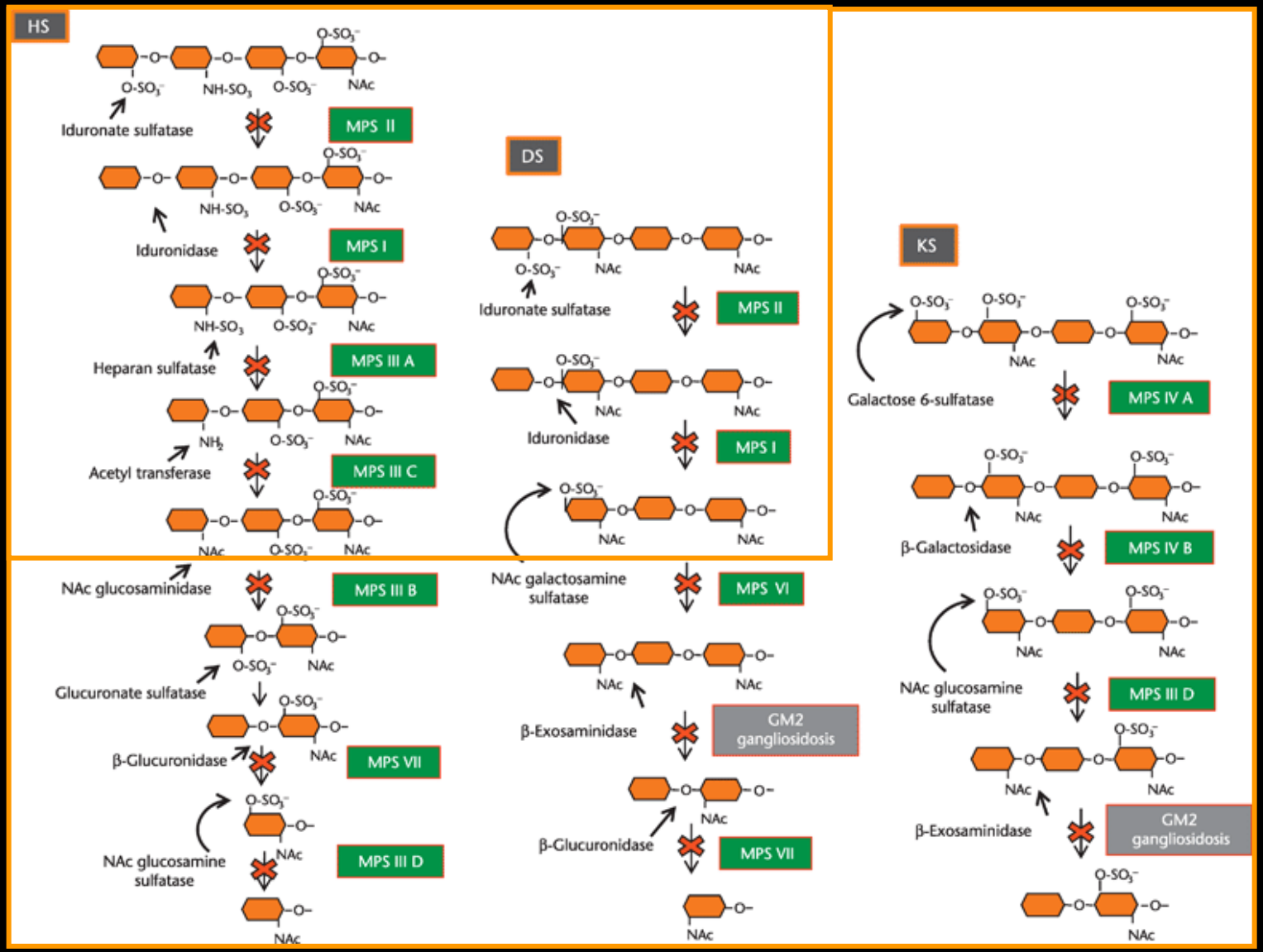
MPS classification

Type	Name	Enzymatic defect	Preval
MPS I	Hurler / Scheie	α-L-iduronidase	1.14
MPS II	Hunter	Iduronate-2-sulphatase	0.74
MPS IIIA	Sanfilippo A	Heparan-N-sulphatase	0.88
MPS IIIB	Sanfilippo B	N-acetyl-α-D-glucosaminidase	0.47
MPS IIIC	Sanfilippo C	Acetyl-CoA α-glucosaminidase	0.07
MPS IIID	Sanfilippo D	N-acetylglucosamine-6-sulphatase	-
MPS IVA	Morquio A	N-acetylgalactosamine-6-sulphatase	0.59
MPS IVB	Morquio B	B-galactosidase	-
MPS VI	Maroteaux – Lamy	Arylsulfatase B	0.43
MPS VII	Sly	B-glucuronidase	0.05
MPS IX	Natowicz	Hyalorunidase	-

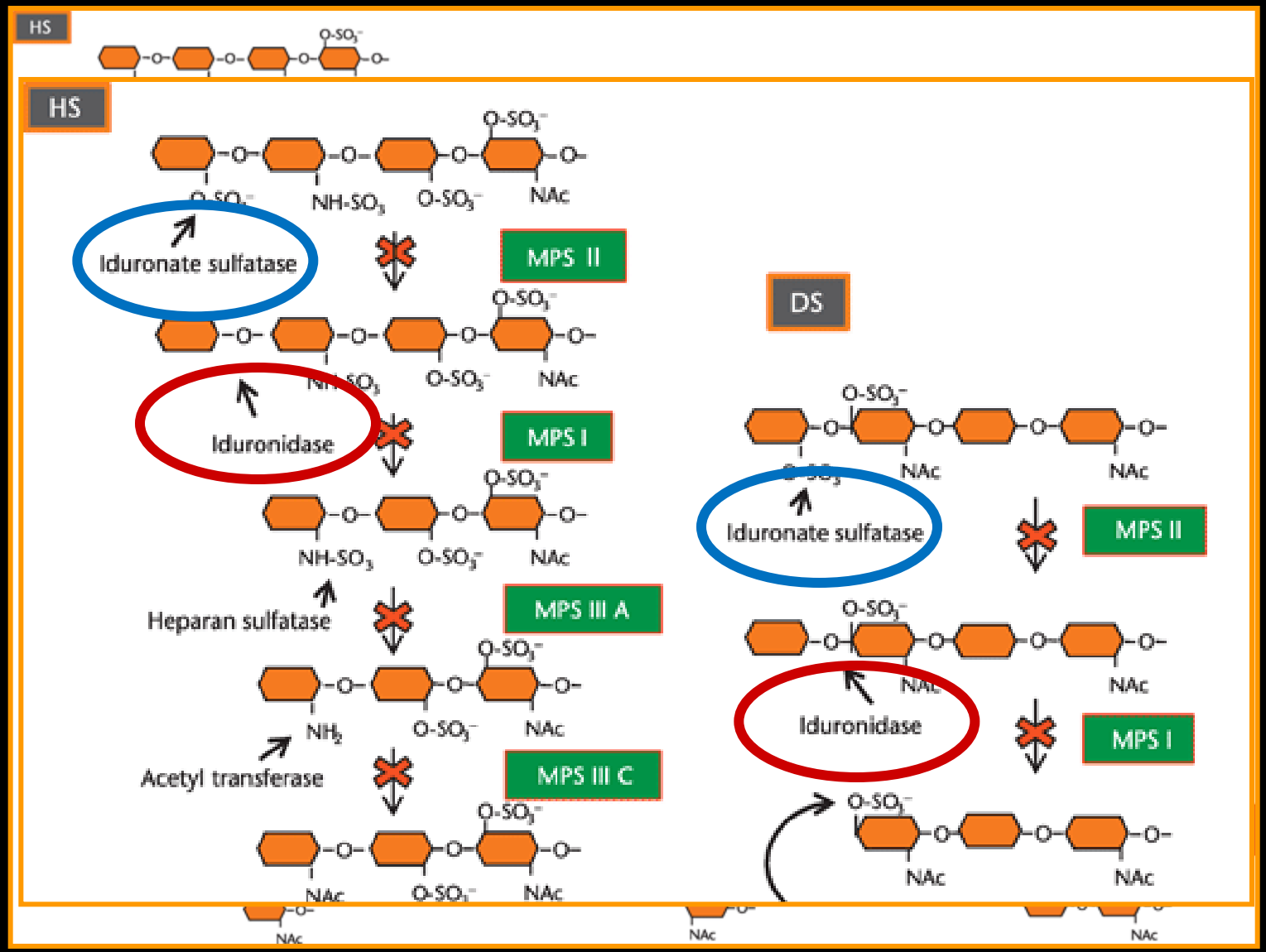
Physiopathology



Physiopathology

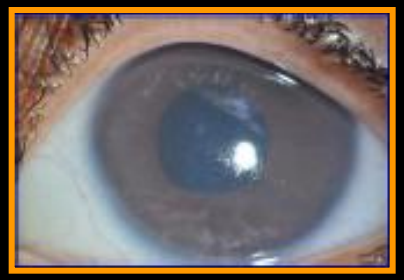


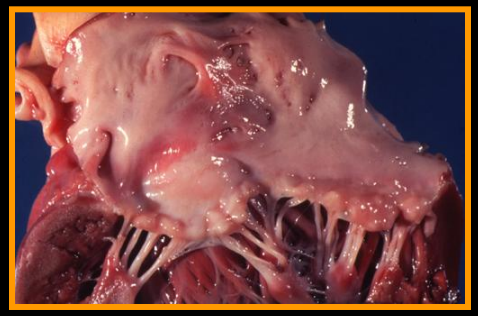
Physiopathology

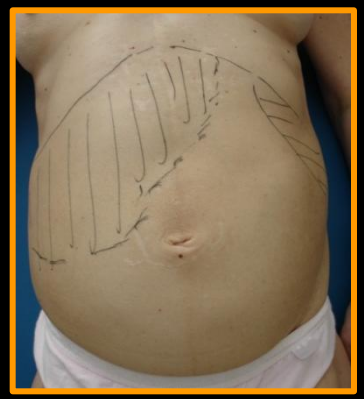


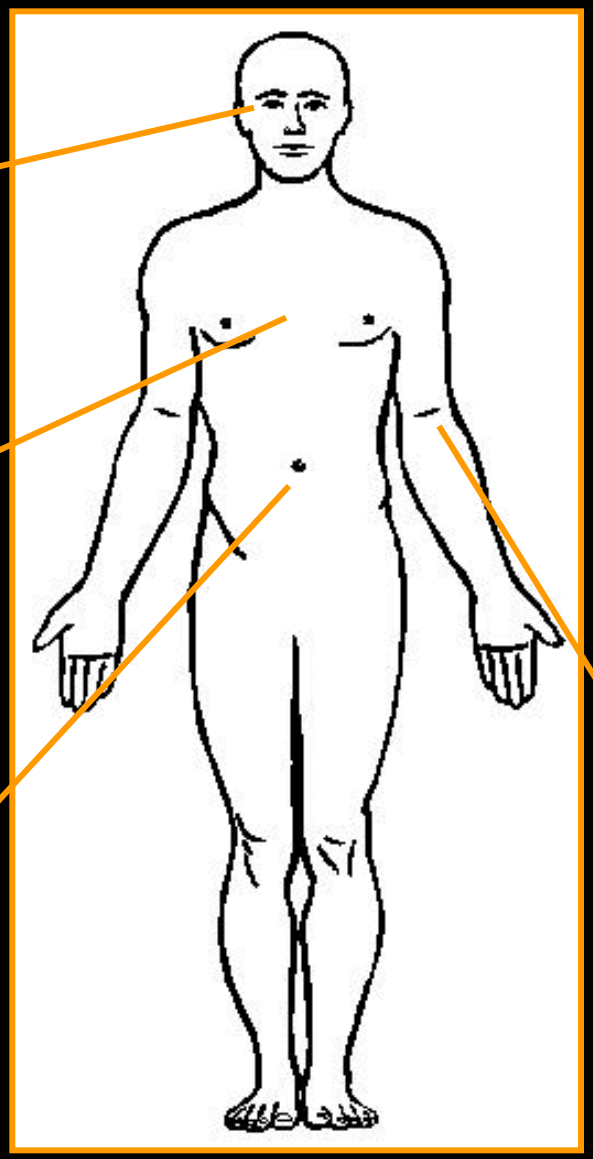
Physiopathology / Clinic

dermatan sulphate

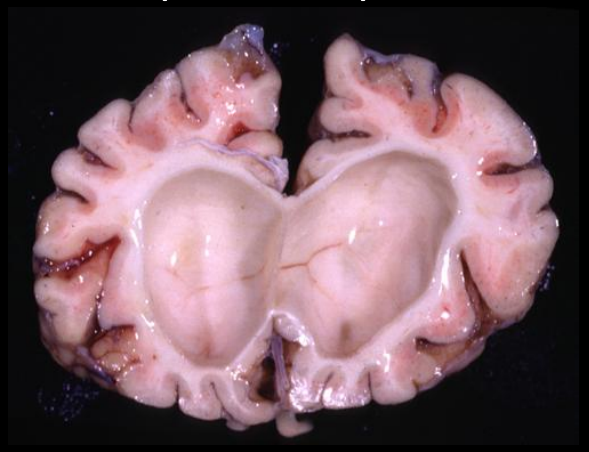




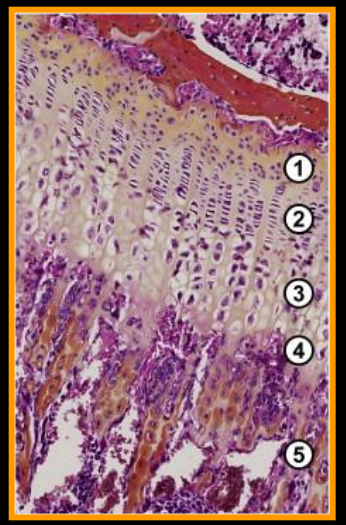




heparan sulphate



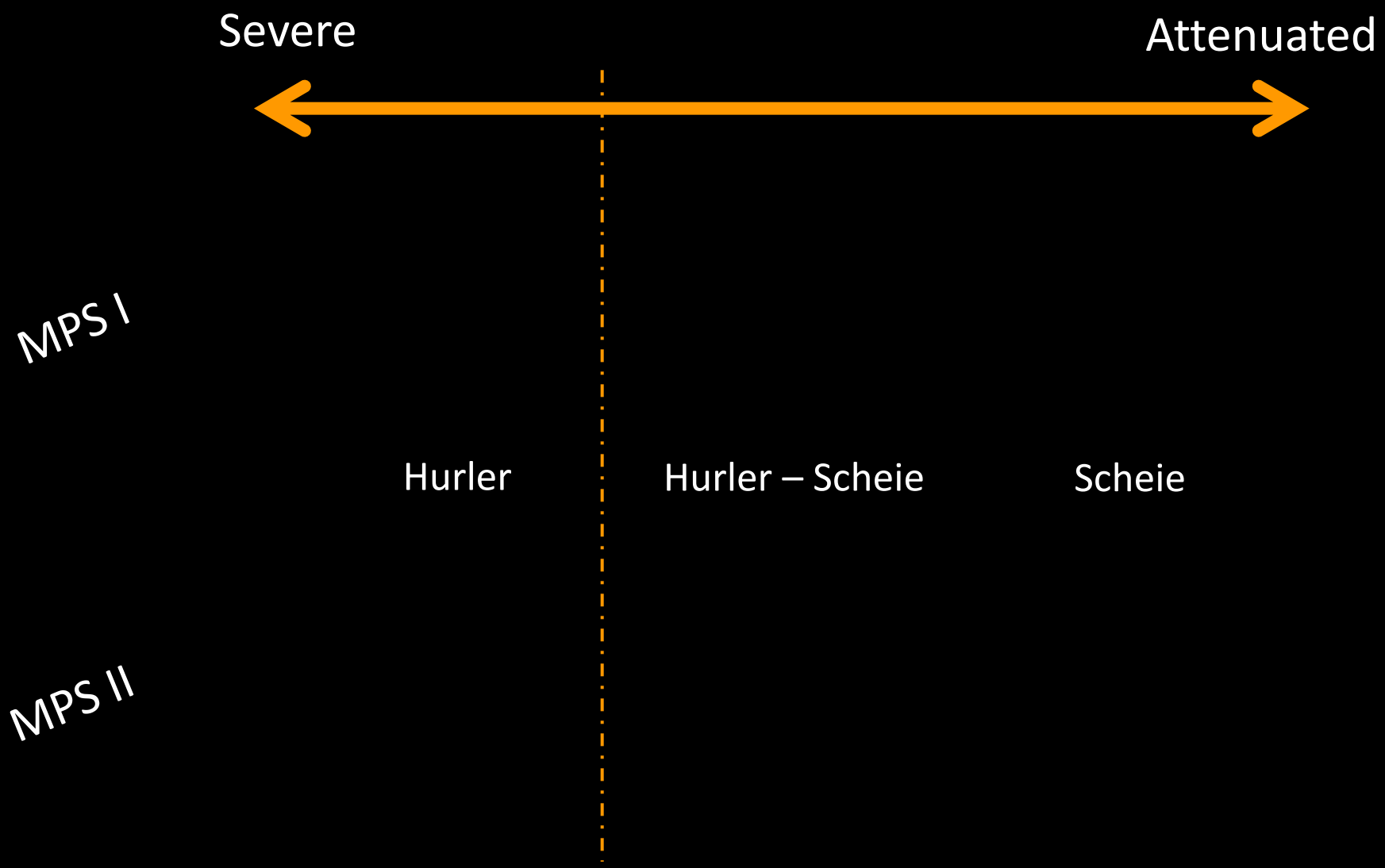
keratan sulphate



Physiopathology / Clinic

GAG	Mucopolysaccharidosis						Clinical manifestations
	I	II	III	IV	VI	VII	
Dermatan sulphate	X	X			X	X	Skeletal and visceral organs
Heparan sulphate	X	X	X			X	Cognitive impairment
Keratan sulphate				X			Skeletal

Clinical spectrum



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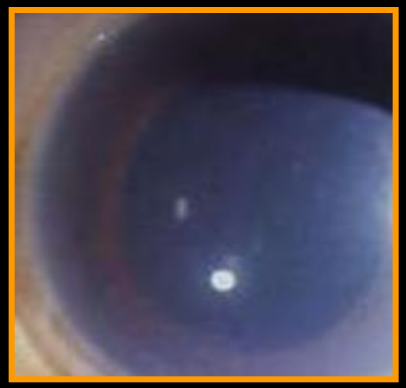
Clinical manifestations

Eye



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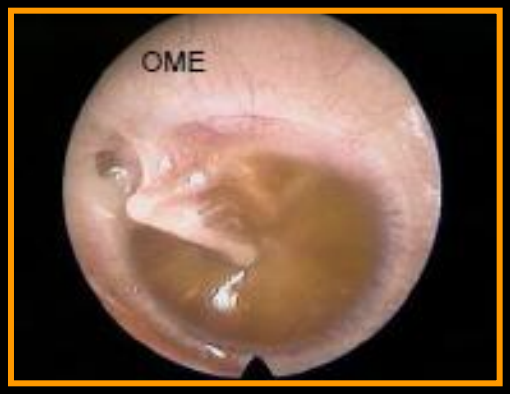
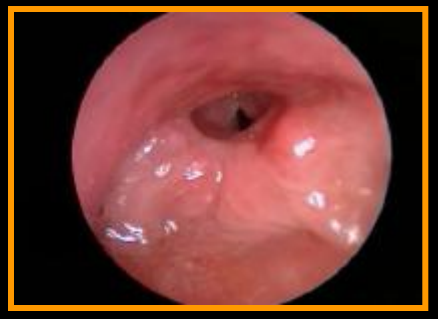
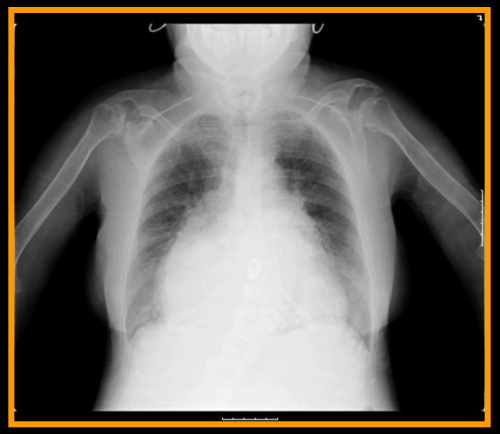


Airway obstruction

(lips to lungs)

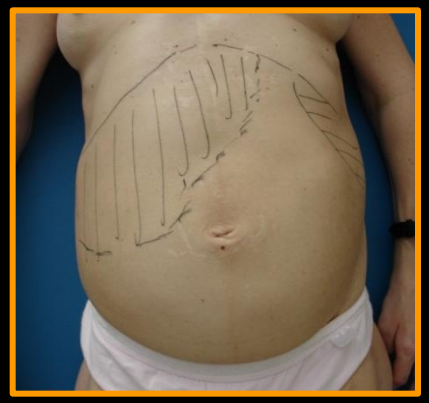


Cardiac

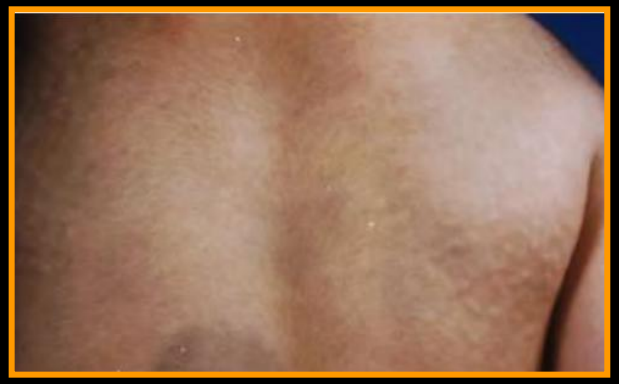


Clinical manifestations

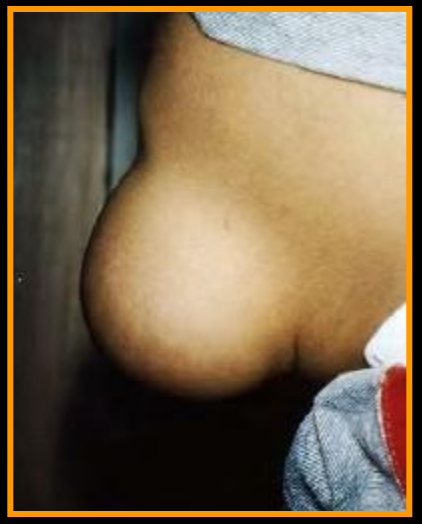
Hepatomegaly



Skin

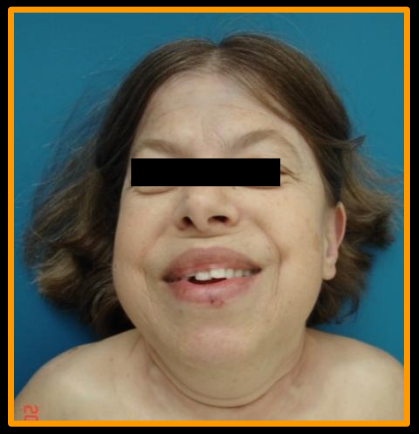


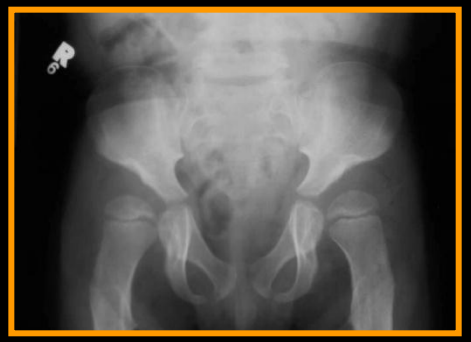
Hernias

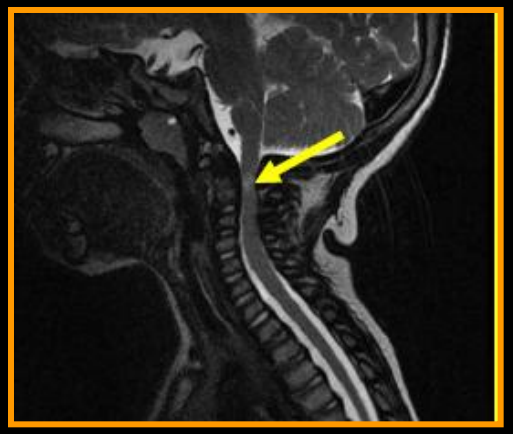


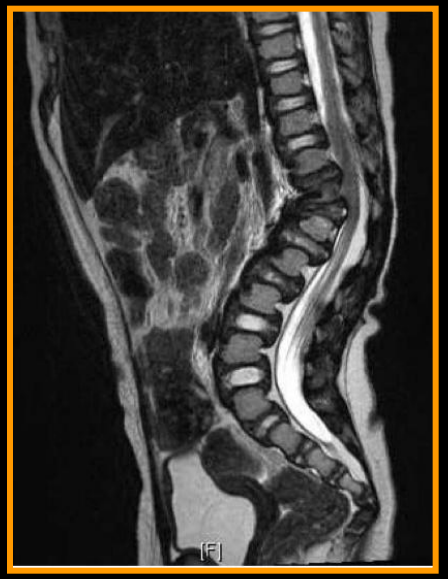
Clinical manifestations

Dysostosis multiplex

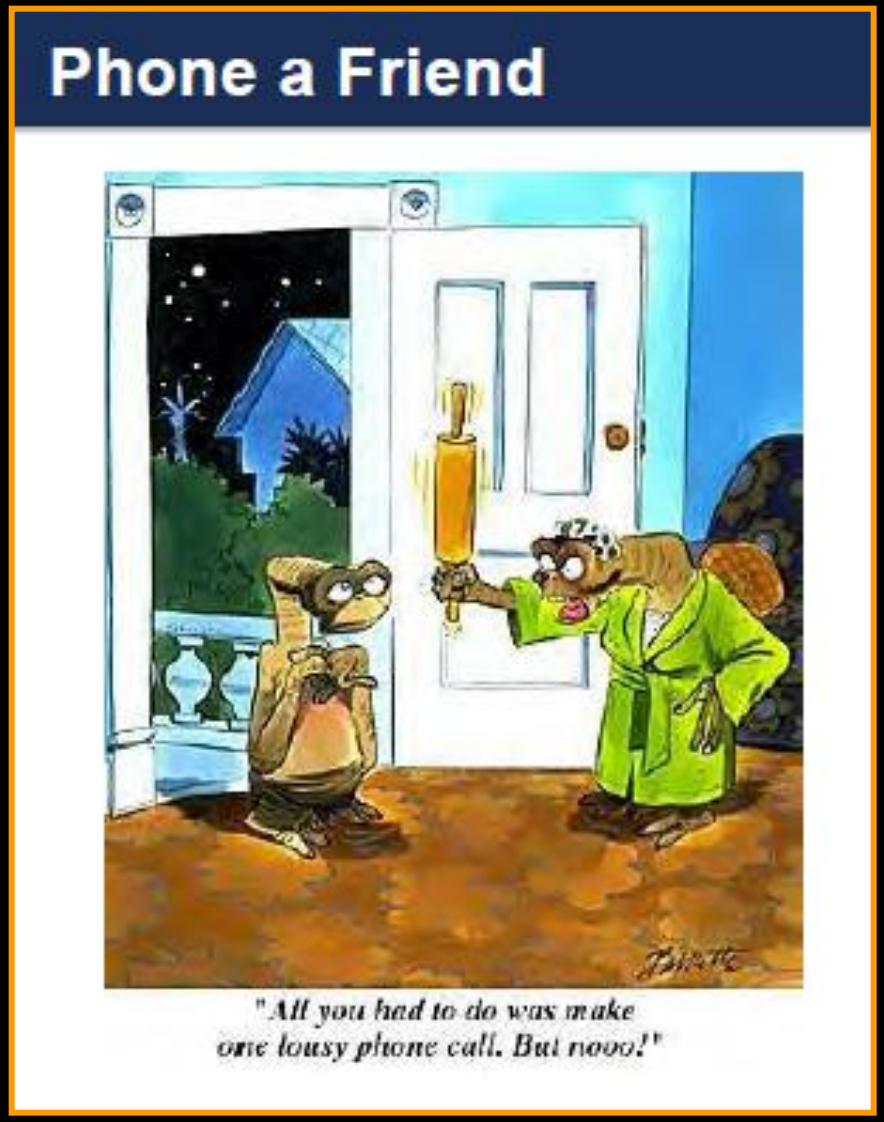






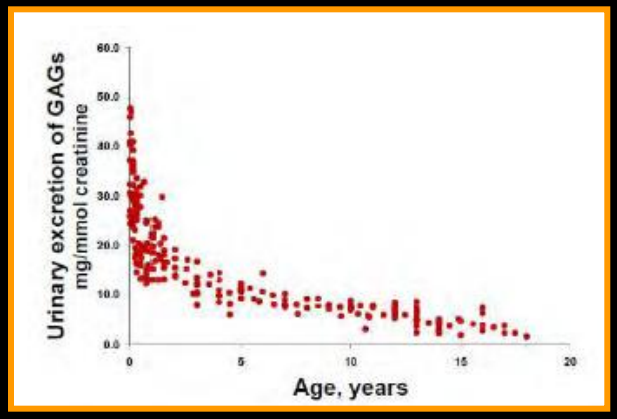


Diagnosis

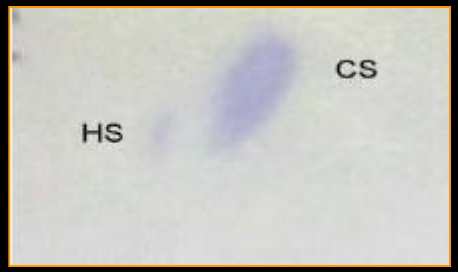

Phone a Friend
"All you had to do was make one lousy phone call. But nooo!"

Diagnosis

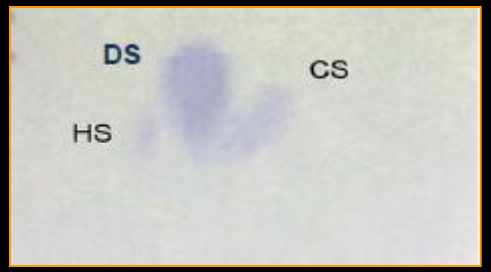
- Urinary GAGs



- Urinary electrophoresis

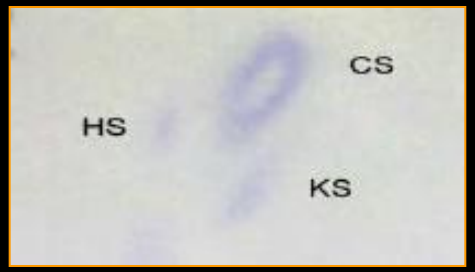


normal



MPS I, II, VI, VII

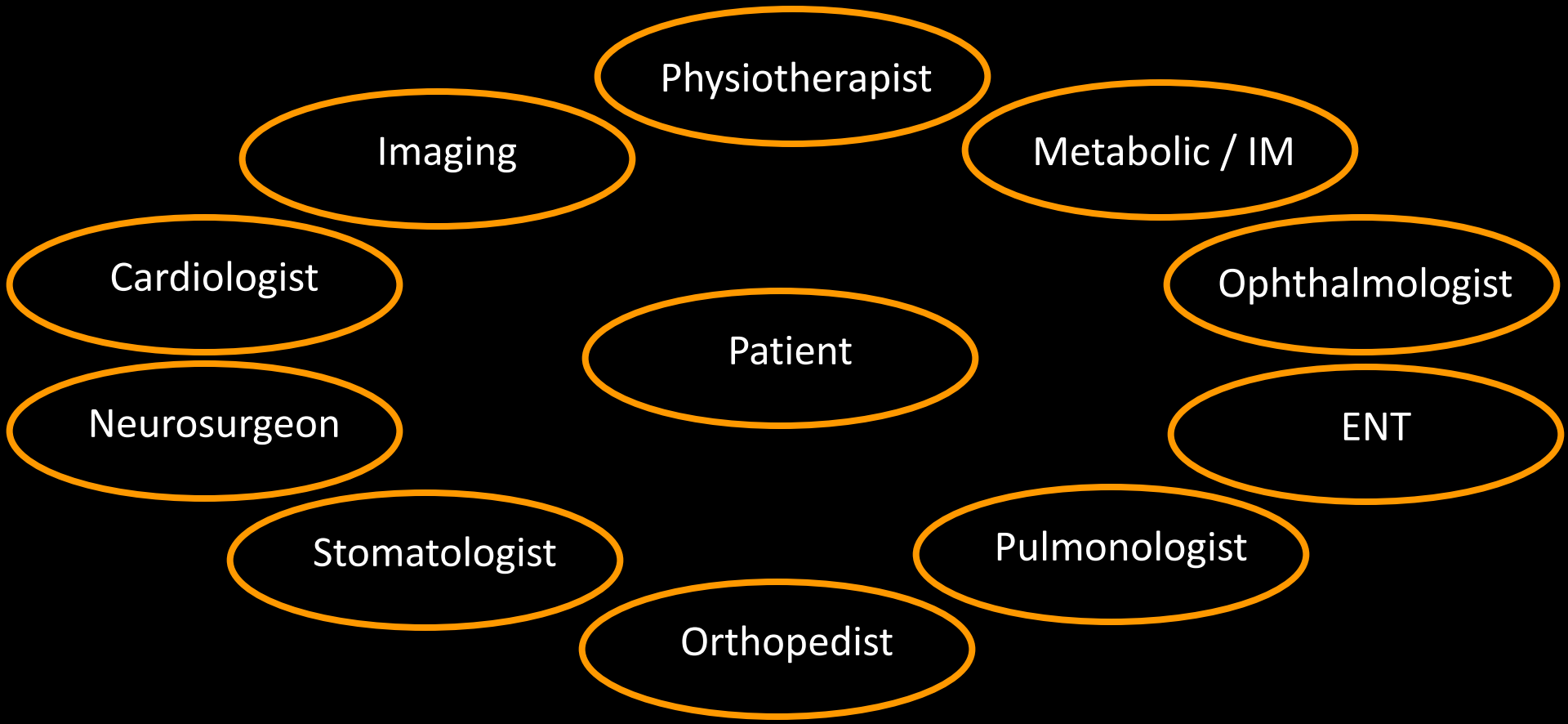
MPS III



MPS IV

- Enzimatic assay
- Gene sequencing

Treatment: multidisciplinary team



Supportive care

Treatment

Type	Therapeutic options
MPS I - H	HSCT
MPS I - HS	HSCT or ERT
MPS I - S	ERT
MPS II severe	ERT or HSCT (?) / clinical trial IT ERT
MPS II attenuated	ERT
MPS III	clinical trial IT ERT
MPS IV	ERT recently approved
MPS VI	ERT
MPS VII	-
MPS IX	-

“There is no disease so rare that it does not deserve attention”

“A disease may be rare but hope should not be”