Ocular Involvement in the Mucopolysaccharidosis: An 8-Year Longitudinal Study

Omar Mehana, MBChB, MSc,[1] , Maha Noor, MBChB,[1,2] Orlaith McGrath[1], Krishanthy Sornalingam, MBBS, MD,[1] MBBCh, Tariq Aslam, DM, PhD,[1,3] Jane Ashworth, BMCh, PhD[1,3].

1. Manchester Royal Eye Hospital, Manchester University Hospitals NHS Foundation Trust, Manchester, United Kingdom 2. Ophthalmology Department, Lancashire Teaching Hospitals NHS Foundation Trust, United Kingdom 3. Manchester University Hospitals NHS Foundation Trust, Manchester, United Kingdom & School of Biological Sciences, Medicine and Health, The University of Manchester, Manchester, United Kingdom.

Purpose

Mucopolysaccharidoses (MPS) are inherited disorders characterized by impaired lysosomal degradation of glycosaminoglycans. Ophthalmic manifestations, including corneal clouding, optic neuropathy, elevated intraocular pressure, and retinopathy in MPS types I, II, III, IV, and VI, can lead to significant visual impairment. This study presents clinical observations and corneal changes in MPS patients.

Method

A prospective study assessed visual acuity, corneal clouding, fundoscopy, OPTOS imaging, optical coherence tomography (OCT), and electroretinography (ERG) in our MPS patients.

Table 2. Outline of Key Demographic Details and Ophthalmic Assessment Findings in the Participants with Mucopolysaccharidosis Recruited in Our Study

Total participants	MPS IH		MPS IH/S		MPS IS		MPS II		MPS IVA		MPS VI	
	29		13		3		9		13		8	
Gender	M	F	M	F	M	F	M	F	M	F	M	F
	14	15	8	5	1	2	8	1	4	9	5	3
Mean age (yrs)	17.79		19.23		30.3		22.78		15		18.125	
Median age (yrs)	17		18		30		17		16		14	
Corneal clouding	None	1 (postbilateral DALK – previously severe)	None	0	None	0	None	7	None	1	None	0
	Mild	13	Mild	8	Mild	2	Mild	2	Mild	12	Mild	2
	Moderate	11	Moderate	3	Moderate	1 (right eye only)*	Moderate	0	Moderate	0	Moderate	4
	Severe	5	Severe	2	Severe	1 *(left eye of same patient)	Severe	0	Severe	0	Severe	2
Median visual acuity (logMAR) (range)	OD: 0.62 (0.08–1.30) OS: 0.60 (0.18–1.20)		OD: 0.20 (0.04–1.06) OS: 0.25 (0.02–1.30)		OD:0.20 (0.20–0.90) OS: 0.52 (0.20–0.84)		OD: 0.10 (−0.18 to 0.30) OS: 0.02 (−0.22 to 0.20)		OD: 0.12 (0.00–0.54) OS: 0.14 (0.00–0.66)		OD: 0.26 (0.00–0.40) OS: 0.28 (0.08–0.60)	
Median IOP (mm Hg) (range)	OD: 21 (7–36) OS: 21 (8–44)		OD: 22 (13–35) OS: 23 (12–56)		OD: 16 (12–22) OS: 14 (11–20)		OD: 20.5 (16–30) OS: 18.5 (16–29)		OD: 17 (9–38) OS: 18 (10–37)		OD: 19.5 (15–34) OS: 22 (9–42)	

DALK = deep anterior lamellar keratoplasty; F = female; IOP = intraocular pressure; logMAR = logarithm of the minimum angle of resolution; M = male; MPS = mucopolysaccharidoses; MPS IH = Hurler syndrome; MPS IH/S = Hurler-Scheie syndrome; MPS IS = Scheie syndrome; MPS II = Hunter syndrome; MPS IVA = Morquio syndrome; MPS VI = Maroteaux Lamy syndrome; OD = right eye; OS = left eye.

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Results

80 MPS patients, aged 3 to 58 years, were evaluated. Various degrees of corneal clouding were present in 71 patients. The cohort included 30 MPS I Hurler, 14 MPS I Hurler-Scheie, 4 MPS I Scheie, 2 MPS II, 13 MPS IV, and 8 MPS VI. Two patients underwent corneal transplants, one had penetrating keratoplasty and the other bilateral deep anterior lamellar keratoplasty (DALK). Median LogMAR visual acuity was 0.62 in patients with MPS I Hurler, 0.18 in MPS I Hurler/Scheie, 0.08 in MPS I Scheie and MPS II, 0.54 in MPS IV, and 0.32 in MPS VI. Retinopathy was found in 32 patients: 25 MPS I, 4 MPS II, 1 MPS IVA, and 2 MPS VI. All patients received enzyme replacement therapy (ERT), hematopoietic stem cell transplantation (HSCT), or both except (2 MPS II & 3 MPS IV).

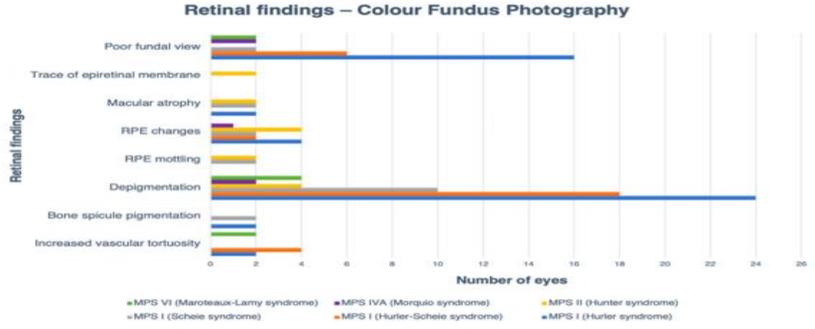


Figure 1. Characterization of retinal abnormalities found on ultrawidefield (UWF) color fundus photography in our participants with mucopolysaccharidosis (MPS) of varying types. RPE = retinal pigment epithelium.

Conclusions

Corneal clouding was prevalent across all MPS subgroups, mostly mild to moderate changes, and 32 patients had retinopathy. While it was challenging to assess progression due to additional ocular conditions, visual acuity remained relatively stable.

References

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