2025 Pathology Update

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Retrospective audit of 3-hydroxy-3-methyl-glutaryl-CoA reductase (HMGCR) antibody testing between 2015 – 2023 in Western Australia

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Appendix 1: Clinical features of patients with positive anti-HMGCR without diagnosis of HMGCR-IMNM

	Year of anti-HMGCR test	Age at symptom onset (years)	Anti-HMGCR antibody level (ref: < 11.4 RU)	Peak CK U/L (ref: 30-170 U/L)	Associated statin use	Clinical history	Lowest grade weakness (MRC scale)	Treatment
1	2016	68	13.4	375	Y	Patient noticed weakness of forearms and then both legs in 2013. Examination Dec 2015: persistent mild weakness of 4+/5 proximally in all limbs. Examination Feb 2016: (Immunology clinic), no proximal weakness	4	Nil
2	2016	21	20.2	1780	N	No symptoms	4	Hydroxychloroquine, sulfasalazine, and prednisolone low dose 5mg intermittently
3	2015	68	11.9	870	Y	Mild facial weakness (orbicularis oculi and oris). Neck flexion 4-, neck extensors 5. Biceps, triceps, wrist significant weakness and long finger flexors. Legs - flexor weakness and knee extension and ankle dorsiflexor weakness. Difficulty swallowing. Diagnosed with inclusion body myositis on muscle biopsy, and positive anti-C5N1A antibody.	4-	Nil
4	2021	39	11	625	N	Nil weakness on Rheumatology clinic review x2 2023. CK felt to be elevated due to ethnicity (Nigerian). Anti-HMGCR antibody repeated 6/10/2023 - negative (3.2 RU).	5	Nil
5	2021	58	24.9	5100	Y	CK noted to be 1550 in 2021; atorvastatin withheld. Hip flexion bilaterally 4/5 suspected secondary to effort. No features of rhabdomyolysis.	4	Nil
6	2022	53	18.7	80	N	SAE1 antibodies (strong positive) detected x3 occasions. Phenotype in keeping with dermatomyositis (DM) typically associated with anti-SAE antibodies (Gottron's papules, heliotrope rash, periungual erythema/capillary loops) and dysphagia with muscle involvement. Previous skin biopsy with features felt to favour DM. There was no IMNM picture clinically.	4+	Prednisolone, mycophenolate, IVIG

Abbreviations:
HMGCR: 3-hydroxy-3-methylglutaryl-CoA-reductase; IMNM: immune-mediated necrotising myopathy; CK: creatine kinase; RU: relative units; MRC: Medical Research Council (a commonly used cale for assessing muscle strength from Grade 5 (normal); C5N1A: cytosolic 5'-nucleotidase 1A; SAE: small ubiquitin-like modifier activating enzyme; DM: dermatomyositis

References
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