Canary in the Carpal Tunnel: A Study of Prevalence and Pilot of a Referral Pathway for Amyloidosis in Carpal Tunnel Syndrome

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INTRODUCTION:

Transthyretin (ATTR) amyloidosis, a form of systemic amyloidosis, can manifest as familial amyloid cardiomyopathy affecting the endomyocardium of the heart. This can result in restrictive cardiomyopathy, and ultimately, heart failure with preserved ejection fraction (HFpEF). Patients presenting with this type of heart failure are often too late for treatment with current medication regimens, and early detection could increase patient lifespan. These misfolded amyloid proteins can also accumulate in the carpal tunnel, leading to carpal tunnel syndrome. This may be the earliest presentation of patients with amyloidosis.

In this pilot study, we propose that early detection of amyloid via a biopsy of the transverse carpal ligament or tenosynovium in the carpal tunnel will allow early detection of systemic amyloidosis and referral to cardiology clinic for consideration of disease-modifying treatment.

METHODS:

Patients undergoing carpal tunnel release who meet criteria for biopsy, as described by Donnelly et al. in 2019, are included. Qualifying patients meet two tier 1 criteria - males aged 50 years or older, females aged 60 years or older, bilateral carpal tunnel symptoms or prior carpal tunnel release surgery; or one tier 1 criteria and one tier 2 criteria - spinal stenosis, biceps tendon rupture, atrial fibrillation or flutter (active or previous history), presence of a pacemaker, congestive heart failure, or family history of ATTR amyloidosis.

Tenosynovial biopsies are taken during open or endoscopic carpal tunnel release and are sent to pathology for congo red staining. If positive, patients are referred to high risk cardiology for additional evaluation and follow up for monitoring and treatment of cardiac involvement.

RESULTS: Of 28 patients matching eligibility criteria screened to date, 4 have had positive biopsy results and have been referred to high-risk cardiology. Of the 4 patients referred, one is currently undergoing advanced cardiac workup for consideration of initiating disease-modifying therapy.

DISCUSSION AND CONCLUSION: This study demonstrates a potential service line link between orthopedic surgery and cardiology. Hand surgeons could potentially be a first point of contact and first line for early diagnosis of transthyretin amyloidosis during carpal tunnel surgeries. This early diagnosis and interdisciplinary treatment pathway could allow for treatment to alter the disease's course and improve quality of life for patients with cardiac amyloidosis.