CARDIOLOGIST **SPECIALIST GUIDE**

Cardiac involvement is present in nearly all MPS II patients and is a major cause of death in this population.[1,2] Cardiac disease can present as early as 5 years of age and most patients exhibit at least one cardiovascular sign or symptom by the second decade of life.[1,3]

Kingston, age 8

Pathophysiology

There is primary glycosaminoglycan storage in the cardiac tissue, mainly in the activated valvular interstitial cells.[4] These cells are normally involved in maintaining valvular structure and integrity and mediating response to injury, however, in MPS II these cells appear to be engaged in attempted, but ineffective, valve repair.[4]

Clinical Consequences

Congestive heart failure, systemic hypertension, valve disease, arrhythmia, and instances of sudden cardiovascular collapse are commonly seen as a result of the structural and functional cardiac abnormalities in MPS II patients.[3,5] Mitral regurgitation is the most common valvular disease in MPS II for the severe phenotype.[5] Before any surgery, a full cardiac assessment is required.[3]

Management

If indicated, regular echocardiography, 12-lead electrocardiography, and Holter monitoring should be considered at the discretion of the cardiologist, typically every 1–3 years, in order to monitor ventricular size and function.[2,5] Hypertension, which is typically underdiagnosed in MPS II patients, may be treated with standard agents, such as angiotensin-converting enzyme inhibitors, angiotensin receptor blockers, diuretics, and calcium-channel blockers.[3]

VALVE DISEASE

CARDIOMYOPATHY

HUNTER SYNDROME IS A MULTISYSTEMIC AND PROGRESSIVE DISEASE

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