**Appendix 2 (as supplied by the authors):** Phenocopies of hypertrophic cardiomyopathy and distinguishing characteristics

| HCM Phenocopies | Differences from hypertrophic cardiomyopathy |
|---|---|
| Athlete's Heart [1, 2] | • Concentric LVH<br>• LVH regresses with cessation of exercise<br>• Cardiopulmonary exercise stress test, Echo parameters of diastolic function may help distinguish from HCM |
| Fabry's Disease[3] | • X-linked recessive multisystem disease<br>o Cardiac manifestations include LVH, conduction defects, CAD, aortic and mitral regurgitation, aortic root dilatation<br>o Renal failure<br>o Stroke<br>o Skin manifestations: telangiectasias, angiokeratomas (typical locations include groin, hip and periumbilical regions)<br>• Diagnosis can be achieved by measurement of low alpha-galactosidase activity, endomyocardial biopsy or genetic testing<br>• Enzyme replacement may alter the disease course |
| Amyloidosis[4] | • ECG is marked by low voltage; Echo may be characterized by LVH with increased echogenicity and decreased systolic function<br>• AL Amyloid<br>o Multisystem disease caused by deposition of monoclonal light chains<br>• ATTR Amyloid; abnormal Tc pyrophosphate scan<br>o Due to autosomal dominant mutations in transthyretin characterized by neuropathy, cardiomyopathy or both<br>o Liver transplantation can be curative, however those with significant cardiac manifestations may require dual organ transplantation |
| Danon Disease[5] | • Characterized by deficiency of lysosomal-associated membrane protein 2 with severe cardiomyopathy and mild skeletal myopathy<br>• Other clinical features can include mental retardation and ophthomologic manifestations<br>• Elevated creatine kinase or muscle biopsy can aid diagnosis |

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|---|---|
| | • Limited treatment options; may progress to need for heart transplantation |
| Mitochondrial Cardiomyopathies[6] | • Characterized by multisystem disease with possible manifestations including:<br>• Optic atrophy; pigmental retinopathy<br>• Cardiomyopathy<br>• CNS features<br>• Peripheral Nervous System features including hypotonia, muscle weakness, neuropathy |
| Noonan Syndrome[7] | • Autosomal dominant disease<br>• Can be characterized by short stature, pectus excavatum, hypertelorism, low set ears, pulmonic valve stenosis amongst other features<br>• Abnormalities platelet function and clotting |

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