

Appendix 3: Table 3: Examples of different diseases that cause cardiomyopathies(Adapted from European Society of Cardiology classification of primary cardiomyopathies¹ (Elliot, et al., 2008).¹

| | HCM | DCM | ARVC | RCM | UNCLASSIFIED |
|-----------------|---|--|---|--|--|
| Familial | Familial, unknown gene Sarcomeric protein mutations β myosin heavy chain Cardiac myosin binding protein C Cardiac troponin I Troponin-T a-tropomyosin Essential myosin light chain Regulatory myosin light chain Cardiac actin a-myosin heavy chain Titin Troponin C Muscle LIM protein <i>Metabolic</i> Glycogen storage disease - Pompe, III, IV, IX - PRKAG2 - Danon Lysosomal storage diseases - Anderson–Fabry - I-cell - Gaucher disease - Aspartylglucosaminuria Sialidosis - Mucopolysaccharidoses (MPS) I, II, III, IV, VI, Mucopolipidoses Fatty Acid Oxidation Defects (FAOD): (particularly if associated with an arrhythmia) - LCHAD and TFP, - VLCAD, CPT2 and CACT deficiency, MADD, - Carnitine deficiency - Selenium deficiency Mitochondrial disorders - 3-methylglutaconic acidurias i.e. Barth syndrome - any mitochondrial respiratory chain defect - Particular mt DNA point mutations (C3303T) - Kearns-Sayre syndrome - Coenzyme Q10 deficiency | Familial, unknown gene Sarcomeric protein mutations (see HCM) Z-band Muscle LIM protein TCAP Cytoskeletal genes Dystrophin Desmin Metavinculin Sarcoglycan complex CRYAB Epicardin Nuclear membrane Lamin A/C Emerin Mildly dilated CM Intercalated disc protein mutations (see ARVC) Mitochondrial cytopathy | Familial, unknown gene Intercalated disc protein mutations Plakoglobin Desmoplakin Plakophilin 2 Desmoglein 2 Desmocollin 2 Cardiac ryanodine receptor (RyR2) Transforming growth factor-b3 (TGfb3) | Familial unknown gene Sarcomeric protein mutations Troponin I (RCM p/2 HCM) Essential light chain of myosin Familial amyloidosis Transthyretin (RCM p neuropathy) Apolipoprotein (RCM p nephropathy) Desminopathy Pseuxanthoma elasticum Haemochromatosis Anderson–Fabry disease Glycogen storage disease | Left ventricular non-compaction Barth syndrome Lamin A/C ZASP a-dystrobrevin |

The electronic version of these guidelines is the version currently in use. Any printed copy cannot be assumed to be current.

| | | | | | |
|------------------------|--|--|---------------|--|---------------------------|
| Familial (cont) | <p>Peroxisomal biogenesis defects</p> <p>Organic acidurias: (associated with metabolic acidosis and frequently associated with arrhythmia)</p> <ul style="list-style-type: none"> - Methylmalonic and propionic acidemia - Disorders of intracellular cobalamin metabolism <p><i>Endocrine</i></p> <ul style="list-style-type: none"> - Congenital lipodystrophies - Congenital generalized lipodystrophy (Berardinelli-Seip syndrome) - Familial partial lipodystrophy <p><i>Syndromic</i></p> <ul style="list-style-type: none"> - Noonan's syndrome - LEOPARD syndrome - Friedreich's ataxia - Beckwith–Wiedemann syndrome - Swyer's-James syndrome (46 XY, pure gonadal dysgenesis) <p>Other</p> <p>Phospholamban promoter</p> <p>Familial amyloid</p> | | | | |
| Non Familial | <p>Obesity</p> <p>Infants of diabetic mothers</p> <p>In utero ritodrine hydrochloride exposure</p> <p>Athletic training</p> <p>Amyloid (AL/prealbumin)</p> <p>Hypertension</p> | <p>Myocarditis (infective/toxic/immune)</p> <ul style="list-style-type: none"> - Viral: Coxsackievirus B, Echovirus, Mumps, Rubella, Rubeola, EBV, Influenza, parainfluenza, Parvovirus - Bacterial: Diphtheria, Meningococci, Pneumococci, Gonococci - Fungal: Candidiasis, Aspergillosis - Protozoal: American trypanosomiasis (Chagas' disease), Toxoplasmosis - Rickettsial: Rocky Mountain spotted fever - Spirochetal: Lyme disease <p>Endocrine</p> <ul style="list-style-type: none"> - Thyrotoxicosis, Hypothyroidism, | Inflammation? | <p>Amyloid (AL/prealbumin)</p> <p>Scleroderma</p> <p>Endomyocardial fibrosis</p> <p>Hypereosinophilic syndrome</p> <p>Idiopathic</p> <p>Chromosomal cause</p> <p>Drugs (serotonin, methysergide, ergotamine, mercurial agents, busulfan)</p> <p>Carcinoid heart disease</p> <p>Metastatic cancers</p> <p>Radiation</p> <p>Drugs (anthracyclines)</p> | Tako Tsubo cardiomyopathy |

The electronic version of these guidelines is the version currently in use. Any printed copy cannot be assumed to be current.

| | | | | | |
|-------------------------------------|--|---|--|--|--|
| Non Familial (cont.) | | <ul style="list-style-type: none"> - Diabetic Cardiomyopathy - Hypoglycemia - Pheochromocytoma - Neuroblastoma - Catecholamine cardiomyopathy <p>Myocarditis (infective/toxic/immune)</p> <ul style="list-style-type: none"> - Viral: Coxsackievirus B, Echovirus, Mumps, Rubella, Rubeola, EBV, Influenza, parainfluenza, Parvovirus - Bacterial: Diphtheria, Meningococci, Pneumococci, Gonococci - Fungal: Candidiasis, Aspergillosis - Protozoal: American trypanosomiasis (Chagas' disease), Toxoplasmosis - Rickettsial: Rocky Mountain spotted fever - Spirochetal: Lyme disease <p>Nutritional</p> <ul style="list-style-type: none"> - thiamine, carnitine deficiency, selenium, hypophosphataemia, hypocalcaemia, Kwashiorkor (protein), Beriberi deficiency, Hypertaurinuria, Vitamin D deficiency <p>Ischaemic</p> <ul style="list-style-type: none"> - Kawasaki disease - ALCAPA - Homozygous familial hypercholesterolaemia and homocysteinuria <p>Tachycardiomyopathy</p> <ul style="list-style-type: none"> - Ectopic atrial tachycardia, PJRT, Slow VT <p>Miscellaneous</p> <ul style="list-style-type: none"> - Eosinophilic (Churg Strauss), Drugs, Alcohol, Peripartum cardiomyopathy, Haemolytic-uremic syndrome, Osteogenesis imperfecta | | | |
|-------------------------------------|--|---|--|--|--|