

INVESTIGATIONS for acute presentation of DCM, HCM, RCM or acute myocarditis

- please keep this list of investigation in the patient notes

Please Note: suggested investigations should be used as guide only and should not supersede clinical judgement

- **Table 1:** First Tier Laboratory Investigations; should be undertaken **immediately** on acute presentation with DCM, acute myocarditis, HCM or RCM
- **Table 2:** Second Tier laboratory investigations; to be undertaken following consultation with the cardiology, metabolic and genetic services. These may be considered when initial investigations do not reveal cause of cardiomyopathy
- Asterisk (*) denotes all blood tests that should be taken pre transfusion or immunoglobulin.
- Immediate referral to metabolic services where metabolic cause of DCM or HCM suspected to expedite tests i.e neonatal cardiomyopathy associated with arrhythmia is highly suggestive of a metabolic cause (i.e long chain fatty acid oxidation defect), contact metabolic consultant immediately.

Blood tests represent ADHB Lab plus requirements and may differ for other laboratories

- Heparin tube - green top, EDTA - lavender/purple top, Plain tube - red top, Fluoride -grey top
- Specimen blood volumes indicated if requesting one test, but can group tests (i.e heparin, plain top and EDTA samples). Contact LAB Link at ADHB ex 5995 for clarification of minimum blood volume needed.

Table 1: First Tier Laboratory Investigations				
Investigation	Specimen Required	Reason	Date	Result (circle if abnormal)
BLOOD: Asterisk (*) denotes blood tests to be taken prior to any transfusion or immunoglobulin				
* Viral studies: Influenza Parainfluenza EBV HIV Parvovirus IgM & IgG	Plain tube: 3mls	Evidence of viral cause of cardiomyopathy		
Enterovirus and parvovirus PCR CMV viral load	EDTA: 0.5ml EDTA: 0.5ml <u>plasma.</u>	Evidence of recent viral infection		
* DNA extraction and storage – mandatory test	EDTA: 3-5mls	DNA stored in case further testing undertaken at a later date		
* Transferrin isoelectric focusing <ul style="list-style-type: none"> • 2nd tier test but do early if considering a blood transfusion 	Plain: 2mls	Congenital disorder of glycosylation (particularly if pericardial effusion and dysmorphism)		
Full blood count	EDTA: 0.5ml	Neutropenia associated with Barth syndrome. Pancytopenia in organic acidaemia		
ESR	EDTA: 0.5mls	Inflammatory marker		
CK	Heparin: 0.5mls	Generalised myopathy of FAOD		
CKMB	Heparin: 0.5mls			
Troponin T	Heparin: 0.5mls			
Electrolytes, urea and creatinine	Heparin: 0.5mls			

Liver function tests <ul style="list-style-type: none"> if AST required order separately 	Heparin: 0.5mls	Abnormal in FAOD, mitochondrial		
CRP	Heparin: 0.5mls	Inflammatory marker		
Thyroid function tests	Heparin: 0.5 ml			
Blood gas (Lactate, Glucose)	Blood gas 0.5 ml	Organic acidaemia		
ANA ds DNA Abs	Plain tube:1ml	investigation and diagnosis of inflammatory connective tissue diseases		
Selenium level	Heparin: 0.5ml	Selenium deficiency		
Red cell transketolase (vitamin B1) <ul style="list-style-type: none"> consult with chemical pathologist prior to requesting 	Heparin: 1.5 ml	Thiamine deficiency		
Vitamin D, Calcium, Phosphate	Heparin: 0.5ml	Vitamin D deficiency		
Acyl-Carnitine Profile <ul style="list-style-type: none"> even if the newborn screening is normal 	Guthrie Card: 1 spot or Heparin: 0.5mls	FAOD, organic acidaemia		
Plasma amino acids	Heparin: 0.5ml	Barth syndrome, Mitochondrial, cobalamin defects		
Homocysteine	EDTA: 1ml (on ice)	Cobalamin defects, homocystinuria		
Alpha glucosidase activity <ul style="list-style-type: none"> Consider as 1st tier test if floppy infant with HCM 	4 spots on Guthrie card	Pompe disease		
STOOL SAMPLE				
For enterovirus <ul style="list-style-type: none"> Prefer stool sample. If stool specimen is unavailable a rectal swab can be sent, but ensure a "pea sized" sample of faecal material is obtained and sent. Request swab and medium from ADHB viral culture Lab 	Stool sample / Rectal swab	Evidence of viral cause of cardiomyopathy		
NASOPHARNGEAL ASPIRATE				
Respiratory Panel PCR; Flu A & B; Adenovirus Parainfluenza 1, 2, 3; CMV; Respiratory syncytical virus, coronavirus, Bocavirus	NPA	Evidence of viral cause of cardiomyopathy		
URINE (NB: can accumulate urine samples)				
Urine organic acids Urine amino acids	Random urine: 3mls	Organic acidaemias, 3-methylglutaconic acidaemias (including Barth syndrome)		
ROUTINE FAMILY SCREENING (for referral to CIDG - download the referral form here)				
Referral to CIDG unless a positive diagnosis of a non familial cause has been made at cidgadmin@adhb.govt.nz				
<ul style="list-style-type: none"> Detailed history – at least 3 generation pedigree ECG/ECHO parents & siblings Further family screening with metabolic and genetic testing may be indicated 		Non familial cause of DCM, HCM and RCM		

Table 2 : Second Tier Laboratory Investigations				
Investigation	Specimen Required	Reason	Date	Result (circle if abnormal)
SPECIALTY GENETIC TESTS: for DCM, HCM and RCM				
DNA testing <ul style="list-style-type: none"> Genetic testing for familial DCM & HCM available Discuss with CIDG 	EDTA: 3-5mls	DNA can be stored in case testing is undertaken at a later date		
Molecular Karotype	EDTA: 4 mls			
SPECIALTY METABOLIC TESTS: for DCM, HCM				
Glucose B-hydroxybutyrate	Heparin: 0.5mls	GSD, mitochondrial		
Lactate	Fluoride: 0.5ml	NB: normal lactate does not exclude mitochondrial disorder		
Alpha glucosidase activity <ul style="list-style-type: none"> Consider immediately (1st tier test) if floppy infant with HCM 	4 spots on Guthrie card	Pompe disease		
Lysosomal white cell enzymes <ul style="list-style-type: none"> call Lab first to confirm collection times 	EDTA: 10mls sent fresh			
Urate	Plain tube: 0.5mls	GSD (glycogen storage disease)		
Lipids	Heparin: 0.5mls	GSD, homozygous FH		
URINE for speciality METABOLIC tests: for DCM and HCM				
Urine oligosaccharides <ul style="list-style-type: none"> discuss with metabolic team first 	Random urine: 5mls	Sialidosis		
Urine glycosaminoglycans	Random urine: 10-20mls	Mucopolysaccharidoses (MPS)		