1. CARDIOVASCULAR

	FISHDGSTIP	Di George syndrome -Tuple 1 probe	**3 ml heparin Bld Heparin**	3 to 7 days
	MD22D	Di George Syndrome-Deletion - 22q deletion	4 ml blood EDTA	3-4 wks
	FISHWS	FISH - Williams syndrome	**3 ml blood Heparin**	3-5 days
	MFHLDLR	Familial Hypercholesterolemia- LDLR gene sequencing	4 ml blood EDTA	6-8 wks
	MFHAPOB	Familial Hypercholesteriemia-APOB gene (exon 26 and exon 29)	4 ml blood EDTA	1-2 wks
	MFHPCSK9	Familial Hypercholesterolemia- PCSK9 gene sequencing	4 ml blood EDTA	6-8 wks
	MFHLP	Familial hypercholesterolemia (Prenatal)	CVS + 4 ml EDTA blood couple/ Affected subject	1-2 wks
	MKCNQ1	Long QT1- KCNQ1 gene sequencing	4 ml blood EDTA	6-8 wks
	MKCNH2	Long QT2- KCNH2 gene sequencing	4 ml blood EDTA	6-8 wks
	MSCN5A	Long QT3- SCN5A gene sequencing	4 ml blood EDTA	6-8 wks
	MLQT	Long QT package (KCNQ1, KCNH2, SCN5A)	4 ml blood EDTA	6-8 wks
	MNKX2	Atrial Septal Defect + Atrioventricular Block- NKX2 gene sequencing	4 ml blood EDTA	6-8 wks
	MML	Marfan syndrome - Linkage studies	4 ml blood EDTA	3-4 wks
	MACADM	MCAD gene sequencing (ACADM gene)	4 ml EDTA	4 to 6 wks
	MCAD	MCAD mutation (Medium chain Acyl-coA dehydrogenase)	4 ml EDTA	2 to 3 wks
	BPOM	Pompe disease - a glucosidase enzyme assay	**6 ml Heparin blood/ DBS**	3-4 days
	MGAAT	Pompe- GAA gene targeted mutation analysis	4 ml blood EDTA	3-4 wks