K. Biochemical and Genetic Studies for Glycogen Storage Disorders

	Inborn Errors of Metabolism – Biochemical Studies		
1	**GSD Screen** (Includes : Glycogen content, Glucose – 6 – Phosphatase (Liver only), Debranching Enzyme, Total Phosphorylase)		
	Liver Muscle		
2	**Glycogen Content And STRUCTURE**		
	Liver Muscle		
3	**Glucose – 6 – Phosphatase (GSD Type Ia)**	GSD I	
	Liver		
4	**Alpha Glucosidase (GSD Type II)**	GSD II	
	Liver, Muscle, Fibroblast, Amniocytes, Chorionic Villi cultures		
5	**Debranching Enzyme (GSD Type III)**	GSD III	
	Liver Muscle		
6	**Branching Enzyme (GSD Type IV)**	GSD IV	
	Liver Muscle Fibroblast		
7	**Phosphorylase (GSD Types V or TypeVI)**	GSD Types V or Type VI	
	Liver Muscle		
8	**Phosphorylase Kinase (GSD Type IX)**	GSD Type IX	
	Liver Blood (RBC) Muscle Heart		
9	**Phosphofructokinase (GSD Type VII)**	GSD Type VII	
	Muscle		
10	**Fructose Assay (**includes: **Fructose 1,6 Diphosphatase & Fructose 1 – Phosphate**		

	Aldolase)		
	Liver		
11	**DNA Test for Glucose – 6 – Phosphatase Gene (GSD Type Ia)**	GSD Type Ia	
	Blood (WBC) Liver Muscle Fibroblast Amniocytes Chorionic villi cells		
12	**DNA Glucose – 6 – Phosphate Translocase (GSD Type Ib)**	GSD Type Ib	
	Blood (WBC) Liver Muscle Fibroblast Amniocytes		
13	**DNA TEST FOR IIIB only** (liver Debranching Enzyme deficiency)		
	Blood (WBC) Liver Fibroblast Amniocytes Chorionic Villi		
14	**LINKAGE STUDY FOR TYPE III (IIIa & IIIb)**		
	Blood (WBC) Liver Muscle Fibroblast Amniocytes Corionic Villi		
15	**DNA Test for Myophosphosrylase Gene (GSD Type V) McArdles**	GSD Type V	
	Blood (WBC) Muscle Fibroblast Amniocytes Chorionic Villi		