

Monogenic Diabetes Study

Criteria for Genetic Testing

PI: Toni Pollin, MS, PhD

Criteria	Description	Intake Questionnaire Indicators	Further investigation (Visit 1)	Further investigation	Genetic Testing
1	Diagnosed with diabetes before the age of 1 year old	# 1 = Yes		IA-2 ab testing. Test C-peptide if on insulin	Yes
2	Diagnosed with T1DM + a parent or child with T1DM	# 5 = Yes AND # 6 = Yes	Test antibodies If IA-2 ab neg	Test C-peptide if on insulin If C-peptide positive	Yes
3a	Diagnosed with non-T1DM at ≤ 30 years old <i>Not on insulin</i>	# 5 = No AND # 2 = Yes	Test antibodies If IA-2 ab neg		Yes
3b	Diagnosed with non-T1DM at ≤ 30 years old On insulin	# 5 = No AND # 2 = Yes	Test antibodies If IA-2 ab neg	Test C- peptide If C-peptide positive	Yes
4	Diagnosed with T2DM at \leq 45 years old + not extremely overweight at diagnosis + two or more relatives with DM diagnosed \leq 50 years old	# 5 = No AND #2 = $Dx \le 45$ AND # 3 = No AND # 7 = Yes If Fam Hx shows 2 or more relatives with DM diagnosed ≤ 50 years old	Pedigree & Medical Hx Test antibodies	If Fam Hx shows 2 or more 1^{st} or 2^{nd} degree relatives (same lineage) with T2DM diagnosed ≤ 50 years old AND If IA-2 ab neg, Test C-peptide if on insulin If C-peptide positive and/or at the discretion of the team	Yes
5	Hearing, visual, or cognitive impairment, birth defects or kidney disease in childhood	# 4 = Yes Team Assessment = Yes to Visit 1	Pedigree & Medical Hx Test antibodies	If IA-2 ab neg Test C-peptide if on insulin If C-peptide positive and/or at the discretion of the team	Yes
6	Non-diabetic with a fasting glucose ≥100 mg/dl during a current or past pregnancy and pre-pregnancy BMI < 25	Gestational DM = Yes AND Confirm documentation of BMI pre- pregnancy (<25) AND glucose during pregnancy \geq 100 mg/dl before scheduling Visit 1	Pedigree & Medical Hx Test antibodies	If IA-2 ab neg Test C-peptide if on insulin If C-peptide positive <i>Or</i> at discretion of team	Yes
7	Clinical suspicion of highly penetrant genetic form of diabetes based on clinician referral or review of available lab results	Complete Intake Questionnaire Team Assessment = Yes to Visit	Pedigree & Medical Hx Test antibodies	If IA-2 ab neg Test C-peptide if on insulin If C-peptide positive and/or at the discretion of the team	Yes



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Antibody Negative = Islet Antigen 2 antibody negative

Positive C-peptide = ≥ 0.6 ng/mL (which converts to ≥ 0.2 nmol/L, depending on units)