

Prader-Willi syndrome DNA (PWS)

Order Name: **PRADR DNA**

Test Number: 5591575

Revision Date: 10/01/2022

TEST NAME		METHODOLOGY	LOINC CODE
Prader-Willi syndrome DNA (PWS)		DNA methylation analysis	

SPECIMEN REQUIREMENTS				
Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	5mL (3mL)	Whole Blood	EDTA (Lavender Top)	Room Temperature

GENERAL INFORMATION	
Testing Schedule	Mon-Fri
Expected TAT	1-2 Weeks
Clinical Use	Prader-Willi syndrome (PWS) is characterized by neonatal hypotonia and failure to thrive, early childhood-onset hyperphagia with resulting obesity, short stature, small hands and feet, hypogonadotropic hypogonadism and mental retardation. The majority of patients (70%) have interstitial deletions of the paternal chromosome 15 (q11.2-q13). Approximately 26% have maternal uniparental disomy (UPD), 2% have chromosome 15 translocations, and 2% have mutations of the imprint control region.
Notes	Prader-Willi syndrome (PWS) DNA methylation analysis can be included in the Hypotonia Panel with myotonic dystrophy (DM) and Spinal muscular atrophy (SMA) analysis to expedite diagnosis. Please submit Pre-Authorization form when the patient has United Healthcare insurance.
CPT Code(s)	(Pre-Authorization Required) 81331
Lab Section	Reference Lab