

RARE DISORDERS PROGRAM

- GeneDx was founded to help patients and clinicians find a diagnosis for patients with rare and ultra rare genetic disorders
- GeneDx has leveraged our experienced staff to build a vast comprehensive test menu based on scientific evidence to broaden our disease offerings

NEW TEST OPTIONS

Disorder Group	Test Code	Test Name	# of Genes
Dysmorphology and Multiple Congenital Anomalies	TA46	Adams-Oliver Syndrome Panel	6
	TA44	Baraitser-Winter Syndrome Panel	2
	T993	Coffin-Siris Syndrome Panel	8
	TA06	Noonan and Comprehensive RASopathies Panel	25
	TA39	Robinow Syndrome Panel	4
	TA38	Treacher Collins Syndrome Panel	6
Hematologic Disorders	TB47	Dyskeratosis Congenita Panel	12
Immunologic Disorders	T990	Autoimmune Lymphoproliferative Syndrome Panel	4
	T989	Chronic Granulomatous Disease Panel	5
	T995	Hyper-IgM Panel	4
Neurologic Disorders	TB51	Comprehensive Holoprosencephaly Panel	17
Pulmonary Disorders	T829	Cystic Fibrosis/Congenital Bilateral Absence of the Vas Deferens single gene test	1
	TB48	Hermansky-Pudlak Syndrome Panel	10
	TB46	Primary Ciliary Dyskinesia Panel	30
	TB49	Surfactant Dysfunction Panel	5
Reproductive Disorders	T991	Neonatal 46, XY Disorders of Sex Development Panel	19
Skeletal Disorders	TA45	Abnormal Mineralization Panel	16
	T992	Autosomal Dominant Osteogenesis Imperfecta Panel	3
	TA40	Craniosynostosis Panel	30
	TA41	Ectrodactyly/Split Hand-Split Foot Panel	13
	T994	Hypophosphatasia and Hypophosphatemic Rickets Panel	9
	TA42	Limb Abnormalities Panel	71
	TA43	Skeletal Dysplasia Panel	29

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In the past few months, GeneDx has expanded our rare test offerings into; immunology, endocrinology, renal, dermatology, pulmonary, dysmorphology, hematology, rheumatology and skeletal disorders.

UPDATED PANELS

Disorder Group	Test Code	Test Name	Previous # of Genes	New # of Genes
Dermatologic Disorders	708	Congenital Ichthyosis XomeDxSlice	39	49
	707	Epidermolysis Bullosa XomeDxSlice	31	28
Dysmorphology and Multiple Congenital Anomalies	584	Cornelia de Lange Syndrome Panel	6	7
Endocrine Disorders	674	MODY Panel	5	16
Hematologic Disorders	J450	Diamond-Blackfan Anemia Panel	Added del/dup testing	
Reproductive Disorders	677	Premature Ovarian Failure Panel	10	22
Skeletal Disorders	T996	Hereditary Multiple Exostoses Panel	2	3
	J797	Osteogenesis Imperfecta Panel	15	24

GeneDx has also added del/dup analysis to over 85 single gene tests!

All rare disorder tests can be ordered from the new Rare Test Requisition Form.
Visit www.genedx.com to learn more!