PEROXISOMAL DISORDERS GENE TABLE

Note: This panel can be ordered from the Mitochondrial/Metabolic test requisition form.

Gene Name	OMIM #	Associated Disorder(s)	Inheritance Pattern
ABCD1	300100	X-linked adrenoleukodystrophy	X-linked
ACOX1	264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
AGPS	600121	Rhizomelic chondrodysplasia punctata type 3	Autosomal recessive
AGXT	259900	Primary hyperoxaluria	Autosomal recessive
AMACR	614307	Alpha-methylacyl-CoA racemase deficiency	Autosomal recessive
DNM1L	614388	*DNM1L*-associated leukoencephalopathy	Autosomal dominant and Autosomal recessive
GNPAT	222765	Rhizomelic chondrodysplasia punctata type 2	Autosomal recessive
HSD17B4	601860	D-bifunctional protein deficiency; Perrault syndrome 1	Autosomal recessive
PEX1	602136	Zellweger spectrum disorder; Heimler syndrome 1	Autosomal recessive
PEX2	614866	Zellweger spectrum disorder	Autosomal recessive
PEX3	617370	Zellweger spectrum disorder	Autosomal recessive
PEX5	600414	Zellweger spectrum disorder; Rhizomelic chondrodysplasia punctata type 5	Autosomal recessive
PEX6	601498	Zellweger spectrum disorder; Heimler syndrome 2	Autosomal recessive
PEX7	614879	Rhizomelic chondrodysplasia punctata type 1	Autosomal recessive
PEX10	614870	Zellweger spectrum disorder	Autosomal recessive
PEX11B	614920	Zellweger spectrum disorder	Autosomal recessive
PEX12	614859	Zellweger spectrum disorder	Autosomal recessive
PEX13	614883	Zellweger spectrum disorder	Autosomal recessive
PEX14	614887	Zellweger spectrum disorder	Autosomal recessive
PEX16	614876	Zellweger spectrum disorder	Autosomal recessive
PEX19	614886	Zellweger spectrum disorder	Autosomal recessive
PEX26	614872	Zellweger spectrum disorder	Autosomal recessive
PHYH	266500	Adult Refsum syndrome	Autosomal recessive
SCP2	604105	Sterol carrier protein deficiency	Autosomal recessive
TRIM37	253250	Mulibrey nanism	Autosomal recessive

207 Perry Parkway
Gaithersburg, MD 20877
T 1 888 729 1206 (Toll-free), 1 301 519 2100 • **F** 1 201 421 2010
E zebras@genedx.com • www.genedx.com

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