

- Thoms S., Erdmann R. 2006. Peroxisomal matrix protein receptor ubiquitination and recycling. *Biochim Biophys Acta* 1763: 1620-1628.
- Walter C., Gootjes J., Mooijer P. A., Portsteffen H., Klein C., Waterham H. R., Barth P. G., Epplen J. T., Kunau W. H., Wanders R. J., Dodd G. 2001. Disorders of peroxisome biogenesis due to mutations in PEX1: phenotypes and PEX1 protein levels. *Am J Hum Genet* 69: 35-48.
- Wanders R. J., Waterham H. R. 2005. Peroxisomal disorders I: biochemistry and genetics of peroxisome biogenesis disorders. *Clin Genet* 67: 107-133.
- Wanders R. J., Waterham H. R. 2006. Biochemistry of mammalian peroxisomes revisited. *Annu Rev Biochem* 75: 295-332.
- Warren D. S., Wolfe B. D., Gould S. J. 2000. Phenotype-genotype relationships in PEX10-deficient peroxisome biogenesis disorder patients. *Hum Mutat* 15: 509-521.
- Weller S., Cajigas I., Morrell J., Obie C., Steel G., Gould S. J., Valle D. 2005. Alternative splicing suggests extended function of PEX26 in peroxisome biogenesis. *Am J Hum Genet* 76: 987-1007.
- Weller S., Gould S. J., Valle D. 2003. Peroxisome biogenesis disorders. *Annu Rev Genomics Hum Genet* 4: 165-211.
- Yahraus T., Braverman N., Dodd G., Kalish J. E., Morrell J. C., Moser H. W., Valle D., Gould S. J. 1996. The peroxisome biogenesis disorder group 4 gene, PXAAA1, encodes a cytoplasmic ATPase required for stability of the PTS1 receptor. *Embo J* 15: 2914-2923.
- Zeharia A., Ebberink M. S., Wanders R. J., Waterham H. R., Gutman A., Nissenkorn A., Korman S. H. 2007. A novel PEX12 mutation identified as the cause of a peroxisomal biogenesis disorder with mild clinical phenotype, mild biochemical abnormalities in fibroblasts and a mosaic catalase immunofluorescence pattern, even at 40 degrees C. *J Hum Genet* 52: 599-606.

Supporting Table S1 (following page): Primer Sequences

Supplemental Table 1. Primer Sequences

Gene	Exon	Forward sequence (5' - 3') ^a	Reverse sequence (5' - 3') ^a	Begin ^b	End ^c
<i>PEX1</i>	1	<u>GAACCCAGAGCGACGCTC</u>	CTGAAGATCAGGTGGCTC	7 ^d	16
	2	GAATCCAGAAAATGTATAC	CAGAGTATAGTCAGCTC	36	142
	3	TACTCCAGAATGGGTGACAG	GCTAAAGACATTGATATTGTG	180	47
	4	TAACTAGATATGGAGTGGAC	CTATAGTGTAGAATATGACAG	41	22
	5a	GCTGTATATTCTACATATAG	<u>CAGAGGTTGCTGACGGTTA</u>	55	421 ^e
	5b	<u>GCAGTCAGGTTGTTCT</u>	GCCACATAAAATTCTCCC	355 ^e	22
	6	TGGACTGAGACAACATGAG	ACTTAGCTAAAGCAACTAGA	126	54
	7	GTAAAGTCAGATATGAGG	GTATGACAGGTTGCAAGAAC	30	59
	8	GTTTCAGACTAACTCTGC	GTTACAAGGAACCTCATAC	23	61
	9	CTGGTATAACAGAACATAGC	GTCTAACATGCTAGTTGGC	81	13
	10	CTGGGAAGGCCAAATTAGC	CCTATATAATAGATGGTC	67	70
	11	GAGGCTGAATCTTGGTG	GCATTATGTATAACATCC	71	50
	12	CGACCTTTCTAGTGGTTTC	GACTAAAAATGCTGACTGAC	68	39
	13	GGTCAACCTTAGAGTATC	GCCTCTAGCACAATATGCAC	79	132
	14	CACTATAGATTTGTCAACCTG	GAAAGAAAGATTCCAAGTTCAAGG	76	19
	15	GTTTCCAGCTAAGATGATGG	CTTCTGGAGTAAGTATTAC	64	21
	16	CTGTTTAGACTTGAGCTC	GAATGCACTAAATGCCAGTG	30	78
	17	GATTCACTTAGACTTAGTCAG	GTTTCAAATATCAGTTCTTC	60	80
	18	CCAACATATGAAGCCTGATT	TCTGATGACATGATGACAT	29	39
	19	GTCATCATGTCATCAGAT	GAGTGTAGCATTGTTG	31	30
	20	GGTACATTCTCAACTTAC	CCATATCCAACTATGGAAC	117	141
	21	TGCCTCAAAACCCTGAGAA	TATGTGGGCTGGTTAGGAG	131	74
	22	CACAGGTATCAGTGAACC	GCTCGTTATAAGTCAAAG	173	72
	23	CCTTAATTAACTCTCGTC	GTAATAGTAGCTGTACTTCC	20	21
	24	GTTCGCCAACAAATCCATTATC	GGTTGAGCGGATAATTAAATG	56	58
<i>PEX6</i>	1a	AACCCCTCAGAGCAAGTTC	<u>CGACTGCTGTCAGGAC</u>	137 ^d	506 ^e
	1b	<u>CGGAGAGACCCCTCCAGT</u>	GGGGCACGAGTCTAAATCT	380 ^e	72
	2	GAGAAGGTTATAACGTGGTG	GAGGGTGAGAAGCTATCCTC	16	22
	3	CATTACTCCCATTGTGACCTG	CACATGATTGCAAGGGAG	24	65
	4	GATTGAGAACCTTGTCTTG	<u>CAGAGGAATCCACTCTCTG</u>	55	120
	5	CAAGGATGGGTAGACCAAG	GATGTTCTAACAGGGCTTTC	54	29
	6	CTAGATCTAAGGGATCTGTG	GGAAATAGCCTGACCCACTG	62	119
	7	GAGGTAGGACTTGGCCA	CAATGTGCTGGATTATAAG	129	61
	8	GCCGACTTGGGACAAGGC	GCAACAGGACTGAGTTCTG	58	54
	9	CTGCTCATGTGCTTCTC	CCATCTACATCCATTCTC	13	32
	10a	<u>CTGCCCCAGGATCAAGAAC</u> ^f	<u>CTGAATGGTCTCCAGGATC</u> ^g	129	130
	10b	<u>CAGGATCAAGAACTCAGGTA</u> ^f	<u>CTGAATGGTCTCCAGGATC</u> ^g	121	130
	11	GAGCAACTGCAAGACAGCTC	CACACAATTGACCTCTGG	103	72
	12	GTATGTCATCTGACGGCTC	GAATGATCATGAGTAGCCTG	26	73
	13	GATGCCAGTGTGTATTGTC	GACTGCTGTGAGCTTCTC	88	52
	14	CTGCCCTGAAAGAGAGGTAC	AGGCAGGTATAAGTTCTAG	64	68
	15	<u>CAACAGACCAGATCTCTG</u>	CCTGCATGTTGATGCATC	125	54
	16	<u>CAAGTATGCTCTGTTAGAG</u>	GTAGCTGGGACTCAGGTGCAC	129	52
	17	CTGACCGAGAACCCATTTC	CTCACTCCAACCTTCATGC	57	12
<i>PEX10</i>	1	GACCCAGAGGTCTGGCC	TGACCACACAGGCAGACAC	94 ^d	68
	2	GAGTGAGTGTGGCTCATG	TCCAGGAGCTTCTCACACTG	109	80
	3	GGGCTCAAGGGCGACAAGATGG	GCATGCACCCAAGTCCAGTG	103	51
	4	GTTCTAACGTGCTCTGTG	CTTACAGGTCTTGTGAAG	85	44
	5	CTTCACAAAGGACCTGTAAAGG	CTCAAAACTGGAGGGTGCTC	36	64
	6	GGTCATTAGTTGAAGGAAATAC	GTGGCTGAGTCCTACCAAGG	120	289 ^e
<i>PEX12</i>	1a	GTCATAGAGTTCTAGTC	<u>CGAGGGTAGTCTCAAGAAC</u>	106	335 ^e
	1b	<u>TGCAGCATTTGACAAGTTAC</u>	CTAGGCTACCAAATAAGCAC	251 ^d	62
	2	GGTGTAGAACCTGTGAAATG	GCCACAAAGTTAACAGGGAG	35	74
	3	GTCACTTGTAATGATACC	<u>AGGCTGAGAAGTGTCAACTC</u>	46	474 ^e
<i>PEX26</i>	2	TTCGCCCAGGCAACT	CGAGGTGTAAGCTCTTG	74 ^d	91
	3	GCTGGAGAGGAACAGTCCC	GATGAAAATCCTCTGTTTC	67	35
	4	GGATAGGAGAACATAGTG	GTTTCACGACAGACCCGAGG	72	56
	5	CTTAGAAAGCAAGCACAGAGG	GTAGGACTCTGACAAACACC	62	70
	6	GATGGCAGAGTGCACAGGTGA	GCCAGGCAGGTTTGAAC	41	30

^asequences in regular type are intronic while those underlined reside within flanking exons unless otherwise indicated^bnumber of bases upstream 3'-base of the forward primer is from the 5'-base of the indicated exon^cnumber of bases downstream the 3'-base of the reverse primer is from the 3'-base of the indicated exon^dnumber of bases upstream the last base of the forward primer is from the ATG start codon^enumber of bases downstream of the first base of the indicated exon^fsequences reside in exon 9 of *PEX6*^gsequences reside in exon 11 of *PEX6*