Supplementary Data: Novel *PEX11B* mutations extend the peroxisome biogenesis disorder 14B phenotypic spectrum and underscore congenital cataract as an early feature

Supplementary Data A: Oligonucleotides used for *PEX11B* analysis

PEX11B sequencing primers:-

Exon 1 L: GTAGCGCGACGGCCAGTCTTCAGTGCTCAGAGCCAAG

Exon 1 R: CAGGGCGCAGCGATGACATGCTCCTCATTCCATCACC

Exon 2 L: GTAGCGCGACGGCCAGTTCTGTCTGTGCCTTTTGGAAG

Exon 2 R: CAGGGCGCAGCGATGACGGAAGGAGGGACAAAGGAAG

Exon 3 L: GTAGCGCGACGGCCAGTCTCAACTTTTGCCCTCTTGC

Exon 3 R: CAGGGCGCAGCGATGACAAGTGATTGGAAGCCAAGTG

Exon 4 L: GTAGCGCGACGGCCAGTTGGAGGACATTTCTAATAGGGG

Exon 4 R: CAGGGCGCAGCGATGACATTCGAATGAGGCTGGCAC

- Underlined bases are N13 tags to facilitate sequencing

PEX11B ddPCR primers:-

Exon 1 L: TGACTAGGGGCGGGAAGT

Exon 1 R: GGCTTGGCTCTGAGCACT

Used together with UPL probe #26

Exon 2 L: TTTTATCCCCCAACCTTTCC

Exon 2 R: GTGCAGAGCCAAATGAGAAA

Used together with UPL probe #12

Exon 3 L: CCTGTGACAATGTCCTGTGG

Exon 3 R: GGGCCCACTTCTCCTGAT

Used together with UPL probe #36

Exon 4 L: ACTCCAGGAGGAGGTCTGC

Exon 4 R: AGCCAGGAGCAGGACTTG

Used together with UPL probe #21

PEX11B dosage PCR primers:-

PEX11B_Ex01dosF: agctaagcgcgagaaggcCCGCTTCAGTGCTCAGAG

PEX11B_Ex01dosR: gctttaccgctcaaccgttGAAAGGAATCATCTCAACAGC

PEX11B_Ex03dosF: agctaagcgcgagaaggcTCACTGTTAGTCACCTCAATC

PEX11B_Ex03dosR: gctttaccgctcaaccgttCACTTCTCCTGATCCACAC

PEX11B_Ex04-1dosF: agctaagcgcgagaaggcCTACTGATGGAGCAAGAGTC

PEX11B_Ex04-1dosR: gctttaccgctcaaccgttGAAGAGATCACAGGCATTTCT

PEX11B_Ex-04-2dosF: agctaagcgcgagaaggcCTCCATCCTGTCTATTCTCAC

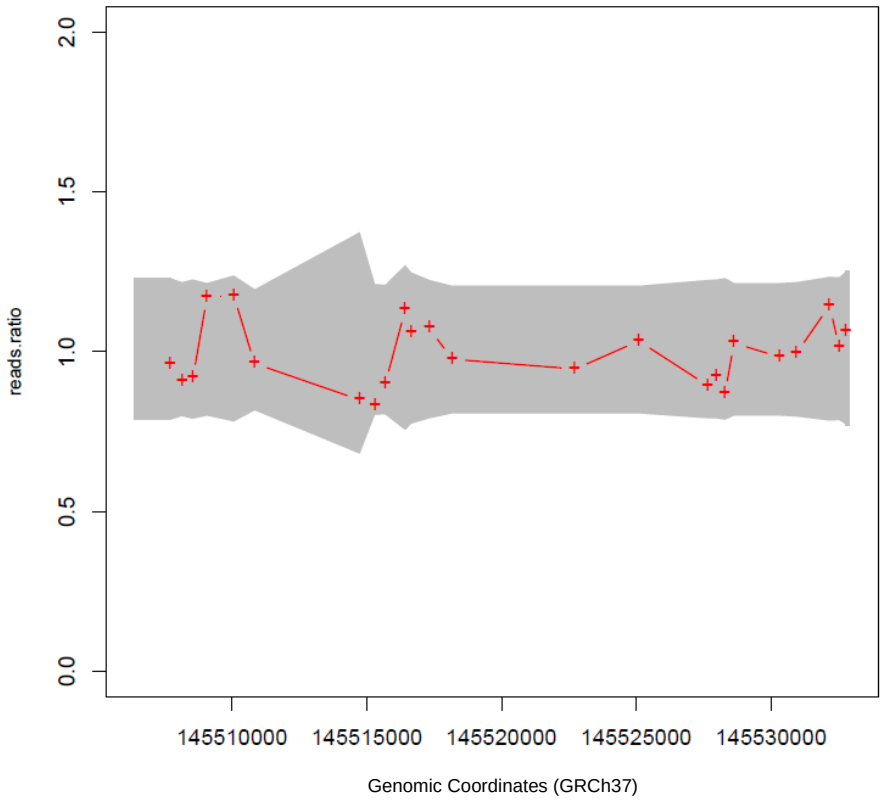
PEX11B_Ex04-2dosR: gctttaccgctcaaccgttGAGTAGAATTCGAATGAGGCT

UniDos_F_FAM: agctaagcgcgagaaggc

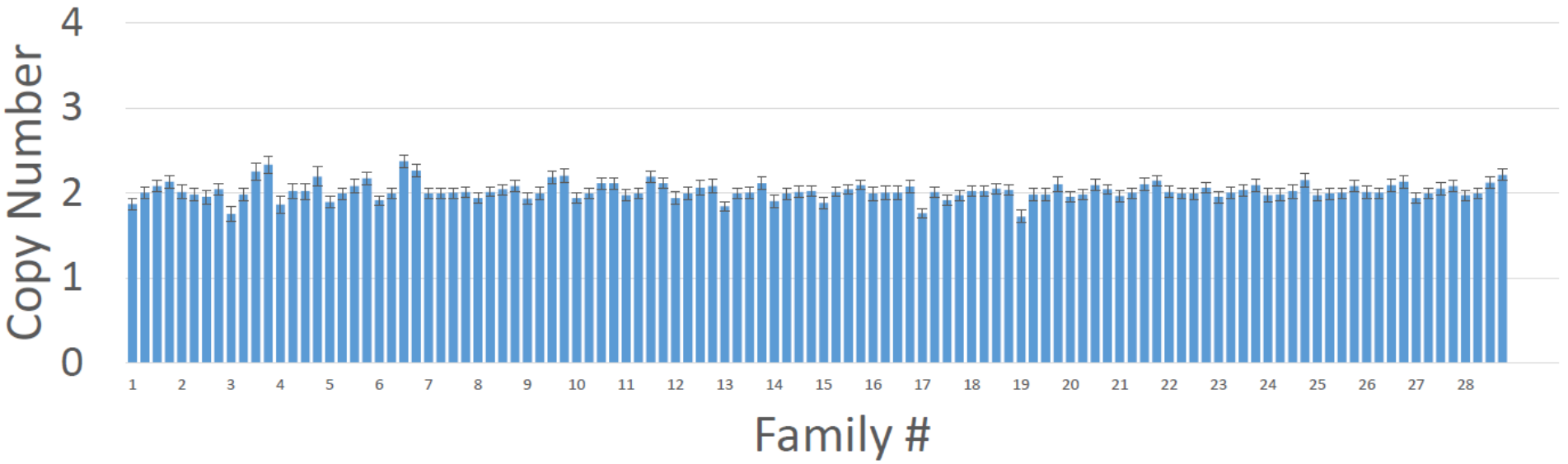
UniDos_R: gctttaccgctcaaccgtt

- Underlined bases are tags to facilitate universal fluorescently labelled primer pair hybridisation

Supplementary Figure S1: ExomeDepth dosage analysis of PEX11B in exome data from patient I.1



Supplementary Figure S2: ddPCR copy number analysis in 28 probands from Warburg micro/Martsolf syndrome cohort.



(# = number)

Supplementary Figure S3: Confirmation of ≥1.87Kbp heterozygous deletion encompassing a minimum of exon 1 to exon 3 of *PEX11B* in Patient III.1 and III.2 by dosage PCR of *PEX11B* ex1-Ex4 (yellow bars) and genomic controls (blue bars)