**Table VI. Diagnostic laboratory studies for PBD**

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| Test | Metabolite/gene | PBD | Comments |
| Plasma VLCFA | Increased C26:0, C26:1 and C24/C22, C26/C22 ratio | ZS, NALD, IRD | False positives in ketogenic diet, non-fasting or hemolyzed samples, normal in RCDP1 |
| Plasma phytanic and pristanic acid | Increased | ZS, NALD, IRDRCDP1 (phytanic only) | Phytanic acid is normal in newborns and accumulates only through dietary intake |
| RBC plasmalogens | Reduced | ZS, NALD, IRD, RCDP1 | Greatest reduction in RCDP1 |
| Urine/plasma bile acid intermediates | Increased di- and tri-hydroxycholestanoic acid | ZS, NALD, IRD | Adjunct to VLCFA and plasmalogen testing |
| Plasma/urine pipecolic acid | Increased | ZS, NALD, IRD | Adjunct to VLCFA and plasmalogen testing |
| Cultured fibroblasts | Confirm abnormal metabolites by enzymatic assays for VLCFAs, plasmalogen biosynthesis and phytanic acid α-oxidation | ZS, NALD, IRD,RCDP1 | Confirmatory testing, also allows more complete characterization of peroxisome functions in difficult cases |
| Molecular genetic testing 1, 2 | PEX 1, 2, 3, 5, 6, 10, 11, 12, 13, 14, 16, 19, 26 | ZS, NALD, IRD | Defects in PEX1, 2, 6, 10, 12, 26 together, account for ~80% of ZSD |
| PEX 7 | RCDP1 | Four common PEX7 alleles in exons 7 and 9, account for ~70% of cases |

1Several *PEX* mutations are more common in Caucasians, due to founder effects:

(1) ~30% of all ZSD alleles are PEX1-Gly843Asp, a missense allele that has residual function. The presence of at least one PEX1-Gly843Asp allele predicts an intermediate or milder (NALD or IRD) phenotype.

(2) ~20% of all ZSD alleles are PEX1-Ile700fs, a frameshift allele, which predicts a severe phenotype when homozygous.

(3) ~50% of all RCDP alleles are PEX7-Leu292X, a nonsense allele, which predicts a severe phenotype when homozygous.

2 Mutation identification is recommended for all patients in order to delineate the mutation spectrum, enhance genotype-phenotype correlations and to better understand disease pathology. It is required when the clinical and biochemical phenotypes do not fit the classic criteria, and for prenatal diagnosis and carrier detection, the latter of which is not possible by existing biochemical methods.