**Supplemental Table S1.** Genetic analysis of patients with HPP

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| --- | --- | --- | --- | --- | --- |
| **Patient** | **Molecular genetic test** | **Exon** | **cDNA-mutation** | **Protein level** | **ACMG** |
| 21001 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 9 | c.984\_986delCTT | p.Phe328del | V |
| 21002 | Compound heterozygous | 6 | c.535G>A | p.Ala179Thr | V |
| 6 | c.571G>A | p.Glu191Lys | V |
| 21003 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 11 | c.1250A>G | p.Asn417Ser | V |
| 21004 | Compound heterozygous | 5 | c.382G>A | p.Val128Me | IV (-V) |
| 11 | c.1276G>A | p.Gly426Ser | III (-IV) |
| 21005 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 10 | c.1001G>A | p.Gly334Asp | V |
| 21006 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 10 | c.1001G>A | p.Gly334Asp | V |
| 21007 | Compound heterozygous | 10 | c.1018C>T | p.His340Tyr | IV |
| 12 | c.1310C>T | p.Ala437Val | III (-IV) |
| 21008 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 12 | c.1354G>A | p.Glu452Lys | IV |
| 21009 | Compound heterozygous | 6 | c.500C>T | p.Thr167Met | V |
| 6 | c.571G>A | p.Glu191Lys | V |
| 21010 | Compound heterozygous | 3 | c.119C>T | p.Ala40Val | IV (-V) |
| 7 | c.746G>T | p.Gly249Val | IV (-V) |
| 21011 | Compound heterozygous | 6 | c.526G>A | p.Ala176Thr | V |
| 10 | c.1114\_1115delCT | p.Leu372Aspfs\*32 | V |
| **Patient** | **Molecular genetic test** | **Exon** | **cDNA-mutation** | **Protein level** | **ACMG** |
| 21012 | Compound heterozygous | 6 | c.526G>A | p.Ala176Thr | V |
| 7 | c.661G>T | p.Gly221Cys | IV (-V) |
| 21013 | Heterozygous | 12 | c.1354G>A | p.Glu452Lys | IV (-V) |
| 21014 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 10 | c.1001G>A | p.Gly334Asp | V |
| 21015 | Compound heterozygous | 5 | c.379A>G | p.Thr127Ala | III (-IV) |
| 6 | c.526G>A | p.Ala176Thr | V |
| 21016 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 10 | c.1001G>A | p.Gly334Asp | V |
| 21017 | Compound heterozygous | 6 | c.526G>A | p.Ala176Thr | V |
| 11 | c.1282C>T | p.Arg428\* | V |
| 21018 | Compound heterozygous | 6 | c.571G>A | p.Glu191Lys | V |
| 12 | c.1354G>A | p.Glu452Lys | IV (-V) |
| 21019 | Compound heterozygous | 10 | c.1009G>A | p.Asp337Asn | IV (-V) |
| 12 | c.1363G>A | p.Gly455Ser | V |
| 21020 | Homozygous | 6 | c.530C>T | p.Ala177Val | IV (-V) |
| 21022 | Compound heterozygous | 6 | c.529G>A | p.Ala177Thr | IV (-V) |
| 7 | c.667C>T | p.Arg223Trp | V |

ACMG, American College of Medical Genetics and Genomics; HPP, hypophosphatasia.