**Table 1.** Gene mutations detected by NGS of two patients.

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| --- | --- | --- | --- | --- | --- |
| **Case** | **Gene** | **cHGVS** | **pHGVS** | **Exon ID** | **Mutation frequency/copy numbers** |
| **case 1** | *CDH1* | c.788C[3>2] | p.Q264Rfs\*18 | EX6 | 58.70% |
| *MEN1* | c.485\_501delTTGGGGCCTGCCAGGCCinsCCA | p.V162Afs\*13 | EX3 | 43.50% |
| *USP6* | c.2021G>A | p.R674K | EX19 | 35.00% |
| *ATRX* | c.1121T>C | p.L374S | EX9 | 2.80% |
| *INPP4B* | c.1834T>A | p.Y612N | EX19 | 1.10% |
| *CCND1* | amplification | NA | all exon | 7.2 |
| **case 2** | *PIK3CA* | c.3140A>G | p.H1047R | EX21 | 14.30% |
| *ERG* | c.277A>T | p.M93L | EX3 | 13.90% |
| *ASXL1* | c.1805delA | p.E602Gfs\*101 | EX13 | 12.50% |
| *FAT1* | c.4592C>T | p.T1531M | EX8 | 1.80% |

ND, not detected; c.HGVS, description of coding DNA (c.) varients by human genome variation society (HGVS); p.HGVS, description of protein (p.) varients by HGVS.

\*describe a stop codon.