

**Supplementary Table S1** Primers sequences for detection *PNKP* mutations

<i>PNKP</i> mutation	Primer sequences, 5'→3'	Amplicon length, b. p.
c.1123G>T, c.1029A>G	F: CGACACGAGGTGCTTCTTGA	798
	R: GAGGACATGAAAGGTCGGG	
c.1270_1283dup14	F: CGGGCACACTGGACGTA	WT-178, mut-192
	R: TGTGACCACGTGTGAGACAG	