Pedigree Symbols

- Normal male
- Normal female
- Affected male or female proband
- Marriage
- Consanguineous marriage
- Diseased
- Sibship
- Twins
Pedigrees of the probands with the variants identified in the present study

SN-68 (proband with NC & variant c.596_597delinsTT in exon 7)

SC-420 (proband with CC & variant c.596_597delinsTT in exon 7)

SN-14 (proband with NC & variant c.422+90 G>A in intron 4)
SN -72 (proband with NC & variant c.422+90 G>A in intron 4)

SN -77 (proband with NC & variant c.422+90 G>A in intron 4)

SP -260 (proband with PSC & variant c.422+90 G>A in intron 4)
SP-272 (proband with PSC & variant c.422+90 G>A in intron 4)

SN -51 (proband with NC & variants c.-979 G>A & c.-738A>G in promoter)

SP -230 (proband with PSC & variant c.422+90 G>A in intron 4)

SP -228 (proband with PSC & variant c.-979 G>A & c.-738A>G in promoter)

SN -55 (proband with NC & variant c.-738A>G in promoter)

SP -263 (proband with PSC & variant c.-471T>G in promoter)
SP -214 (proband with PSC & variant c.-738A>G in promoter)

SP -229 (proband with PSC & variant c.-471T>G in promoter)