Table 1. : Clinical features of three patients with PURA syndrome

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| Mutation | c.692T>G (p..Phe231Cys) heterozygousvariant |  c.159dupG heterozygous variant | c.677\_678deITGheterozygous variant |
| Age, Gender | 3y, male | 13y, male | 4y, female |
| GrowthShort Stature | No | Yes 115 cm (-4,92 SD) | Yes 87 cm (-4,64 SD)  |
| Neonatal ProblemsHypotoniaFeeding DifficultiesRefluxBreathing ProblemsHypersomnolenceHypothermia | YesYesYesYesNoNo | YesYesYesNoNoNo | YesYesNoYesYesNo |
| Neurological AbnormalitiesCognitive ImpairmentHypotoniaStereotypic Hand MovementsEpilepsyMovement DisorderBrain Abnormalities | YesYesNoNoNoNo | YesYesNoYesNoNo | YesYesNoNoNothin corpus callosum |
| Skeletal AbnormalitiesScoliosisHip DysplasiaHyperlaxity | NoNoNo | YesNoYes | NoYesNo |
| Cardiac Abnormalities | No | No | No |
| Urogenital Abnormality | No | No | No |
| Ophthalmological Abnormality | No | Yes | No |