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

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| Mutation | c.692T>G (p..Phe231Cys) heterozygous  variant | c.159dupG heterozygous variant | c.677\_678deITG  heterozygous variant |
| Age, Gender | 3y, male | 13y, male | 4y, female |
| Growth  Short Stature | No | Yes 115 cm (-4,92 SD) | Yes 87 cm (-4,64 SD) |
| Neonatal Problems  Hypotonia  Feeding Difficulties  Reflux  Breathing Problems  Hypersomnolence  Hypothermia | Yes  Yes  Yes  Yes  No  No | Yes  Yes  Yes  No  No  No | Yes  Yes  No  Yes  Yes  No |
| Neurological Abnormalities  Cognitive Impairment  Hypotonia  Stereotypic Hand Movements  Epilepsy  Movement Disorder  Brain Abnormalities | Yes  Yes  No  No  No  No | Yes  Yes  No  Yes  No  No | Yes  Yes  No  No  No  thin corpus callosum |
| Skeletal Abnormalities  Scoliosis  Hip Dysplasia  Hyperlaxity | No  No  No | Yes  No  Yes | No  Yes  No |
| Cardiac Abnormalities | No | No | No |
| Urogenital Abnormality | No | No | No |
| Ophthalmological Abnormality | No | Yes | No |