**Table S1.** Clinical features of 12q21 deletion syndrome

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Brady et al.1999 | Rauen et al.2000 | Rauen et al.2002 | Klein et al.2005 | Present case |
| Cytogenetic Karyotype | 46,XX,del(12)(q21.2q23.2) | 46,XX,del(12)(q21.2q22) | 47,XYY,del(12)(q21.1q21.33) | 46,XY,del(12)(q21.1q22) | 46,XY,del(12)(q21.2q21.33),inv(12)(q13q21) |
| Sex | Female | Female | Male | Male | Male |
| Intellectual disability  | + | + | + | + | + |
| Sparse hair | + | + | + | + | + |
| Prominent forehead | + | + | + | + | + |
| Hypo-/hyper-telorism | Hyper | Hyper | Hypo | Hyper | Hypo |
| Ocular abnormality | − | + | − | + | + |
| Downslanting palpabral fissures | − | + | + | − | − |
| Short upturned nose | − | + | + | + | + |
| Small mandible | No report | + | + | No report | + |
| Low-set ears | + | + | + | + | + |
| Skin findings | No report | Hyperkeratotic eruption | Hyperkeratotic eruption  | Dermatitis | − |
| Cardiac abnormality | − | small VSD | PDA | PDA | − |
| Cranial MRI | No report | VentriculomegalyDelayed myelination | − | Ventriculomegaly | VentriculomegalyHypoplasia of the corpus callosum |

VSD: ventricular septal defect; PDA: Patent ductus arteriosus.