**Supplemental Table 1**. Comparison of clinical and radiological features of previously patients described presenting the typical DBDQ feature with pathogenic variants in *SLC26A2/DTDST* gene and P12 from our cohort.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Phenotype** | | **DD/rMED/DBQD**  Miyake *et al*., 2008 | **DD/DBQD**  Panzer *et al*., 2008 | **Desbuquois-like**  P12 – present cohort |
| ***SLC26A2*/*DTDST* genotype** | | p.[T266I];[V340del] | p.[A133V];[R178\*] | p.[R279W];[A719Qfs\*16] |
| Consanguinity | | - | - | - |
| **At birth** | |  |  |  |
| Weight | | na | 3.310 g (p25-50) | 2,755 g (p75) |
| Length | | 49.5 cm (p25-50) | 47 cm (p3-10) | 41 cm (p3) |
| Occipitofrontal Circumference | | na | 35.5 cm (p75-90) | 33 cm (p75) |
| Gestational Age | | 40 weeks | 40 and 3/7 weeks | 35 weeks |
| **Clinical features** | |  |  |  |
| Unusual facies | | + | + | - |
| Small Stature | | + | + | + |
| Short Limbs | | + | + | + |
| Rhizomelia | | na | + | + |
| Cleft palate | | na | + | - |
| Intrauterine growth retardation | | - | - | + |
| Cognitive disability | | na | - | - |
| Stiff Joints | | + | + | + |
| Laryngomalacia | | na | + | - |
| Cystic ear | | na | + | - |
| Extension camptodactyly | | - | - | - |
| Bilateral clubfoot | | + | unilateral | - |
| Height | | -3SD at 4 yo | na | -3SD 4 yo |
| Hyperextension of bilateral knees | | + | - | + |
| Contracture of the MP joints | | + | - | - |
| **Radiologic features** | |  |  |  |
| Swedish key appearance of the femora | | + | + | + |
| Bilateral hip subluxation/Coxa vara? | | + | - | + |
| Precocious carpal ossification | | + | + | + |
| Brachydactyly | | + | + | - |
| Aberrant ossification centers of wrists at birth | | - | + | - |
| Flat acetabular roof | | ? | + | + |
| Scoliosis | | + mild at 4 yo | + (congenital) | - |
| Extra ossification center of the proximal phalanx of 2nd finger | | - | - | - |
| Ulnar deviation | | na | + | - |
| Cervical kyphosis | | - | + | - |
| Hypoplastic cervical vertebrae | | mild flattening vertebrae | + | + |
| Others | | - | mild macrocephaly | - |
|  |

na: not available; TD: Transmembrane domain; STAS: Sulphate Transporter and Anti-Sigma factor antagonist. EC-loop: Extracellular loop; AO-2: atelosteogenesis type 2; DD: diastrophic dysplasia; rMED: recessive multiple epiphyseal dysplasia; DBQD: desbuquois dysplasia. MP: Metacarpophalangeal