**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