**Supplementary Table 3. Clinical characteristics and demographic features of patients with decreased bone quality**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Abbot et al. 2017[1] | 1 | F | Neonatal (died at day 3) | NM | Homozygous pathogenic mutation in LMOD3 | Congenital fractures of humeri and a fracture of the right femoral diaphysis | Unknown | No |  |
| 2 | F | Neonatal (died at day 43) | NM | Homozygous pathogenic mutation in LMOD3 | Congenital fractures of the right humerus and left femur | Unknown | No |  |
| 3 | F | Neonatal (died at day 9) | NM | Homozygous pathogenic mutation in LMOD3 | Congenital fracture of the left humerus | Unknown | No |  |
| Bharucha et al. 2013[2] | 4 | Unknown | 2y4mo | CCD | RYR1 (ex 12 c.1201C>T p.Arg401Cys; ex 34 c.7463\_7475del13 p.Pro2488HisfsStop39) | Congenital fractures of bilateral femur | Unknown | No |  |
| 5 | Unknown | Neonatal (died at day 24) | CCD | RYR1 (ex 100 c.14422-14424delTTC p.Phe4808del;ex 19 c.2275A>G p.Asn759Asp) | Congenital fractures of left humerus and bilateral femur | Unknown | No |  |
| Böhm et al. 2018[3] | 6 | Unknown | Neonatal (died shortly after birth) | ASCC1-RM | c.157dupG (p.Glu53fs19\*) | Congenital fractures of humeri | Unknown | No |  |
| 7 | Unknown | Neonatal (died shortly after birth) | ASCC1-RM | c.157dupG (p.Glu53fs19\*) | Congenital fractures of humeri) | Unknown | No |  |
| 8 | Unknown | Neonatal (died shortly after birth) | ASCC1-RM | c.157dupG (p.Glu53fs19\*) c.466C>T (p.Arg156\*) | Congenital femoral and humeral fractures | Unknown | No |  |
| 9 | Unknown | Neonatal (died shortly after birth) | ASCC1-RM | c.412C>T (p.Arg138\*) | Multiple congenital bone fractures | Unknown | No |  |
| 10 | Unknown | Neonatal (died shortly after birth) | ASCC1-RM | c.412C>T (p.Arg138\*) | Multiple congenital bone fractures | Unknown | No |  |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Brown et al. 2019[4] | 11 | F | 26 y | NM | Unknown | Fracture of midshaft of left femur after a transfer | Osteope-nia | No | Miniscule diaphyseal size |
| Buonocore et al. 1993[5] | 12 | F | Neonatal (died at 4,5 months) | NM | Unknown | Congenital fracture of both femurs and left humerus | Physiolo-gical remodel-ling of the fractures and no visible osteopo-rosis on X-ray (perfor-med at 2 months) | No | Normal calcium homeostasis |
| Cahill et al. 2007[6] | 13 | Unknown | Paediatric age | MTM | Unknown | 9 fractures | Unknown | Yes (1), No (2) | Particularly metaphyseal fractures, especially of the femora; 1 refracture; fracture healing did not seem to be altered; no operative intervention; long and thin long bones with normal cortical widths |
| 14 | Unknown | Paediatric age | MTM | Unknown | 3 fractures |
| 15 | Unknown | Paediatric age | MTM | Unknown | 3 fractures |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Castiglioni et al. 2014[7] | 16 | M | Neonatal | MTM | MTM1 splicing mutation in intron 5 (IVS5: -1 G>A) | Congenital mid-diaphysis fracture of the left femur and right humerus | Unknown | Unknown |  |
| 17 | M | Neonatal (currently 6 years old) | RYR1-RM | Heterozygous RYR1 mutation | Congenital fractures of the right humerus and left femur | Unknown | Unknown |  |
| 18 | F | Neonatal (died at 3 months) | NM | ACTA1 (de novo mutation, p.G148V (Glycine to Valine) c.443G>T) | Congenital fracture of left humerus | Unknown | No |  |
| 19 | F | Neonatal (died at 2,5 months) | NM | ACTA1 (de novo mutation, c.282C>G, p.N94K (aminoacid change: asparagin to lysine) | Congenital fracture in the third half of the left femur | Unknown | No |  |
| Coppens et al. 2019[8] | 20 | Unknown | Neonatal | Congenital titinopathy | Unknown | Congenital long bone fracture | Unknown | Unknown |  |
| 21 | Unknown | Neonatal | Congenital titinopathy | Unknown | Congenital long bone fracture | Unknown | Unknown |  |
| 22 | Unknown | Neonatal | Congenital titinopathy | Unknown | Congenital long bone fracture | Unknown | Unknown |  |
| Donaldson et al. 1985[9] | 23 | M | Neonatal | Nonspecific neonatal myopathy | Unknown | Congenital fractures of both humeri, both femora and the left tibia | Unknown | Unknown |  |
| Gangfuss et al. 2021[10] | 24 | M | 13 months (age at diagnosis); 11 years (current age) | MTM | c.342 + G>A; p.(Ser79\_Aspl15del) | Unknown | Osteopo-rosis | No |  |
| 25 | M | 4 months (age at diagnosis); died at 2 years | MTM | splice site mutation (c.1261-10A>G, p.(Ser420\_Arg421insPheIleGln) | Fractures of both femurs and multiple ribs without adequate trauma (age 1 year) | Unknown | No | Hypocalcae-mia and hypophospha-temia; vitamin D and calcium supplementa-tion |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Garcia et al. 2009[11] | 26 | M | Neonatal (died at 2 months) | NM (ACTA1) | Two heterozygous missense mutations in exon 3 (g.2007G > T (c.222G > T) and g.2008C > T (c.223C > T)) | Congenital fractures of both femurs and the right arm | Unknown | No | Surgical treatment of the fractures |
| Ikeda et al. 2010[12] | 27 | M | 14 years (presentation: neonatal) | X-linked myopathy with excessive autophagy associated with VMA21 mutation | c.164-6T>G substitution in intron 2 in VMA21 | Not described | Osteopo-rosis (BMD was extremely reduced) | Ambulant at age 2 years; wheelchair bound by age 5 years | Bone turnover markers were elevated |
| 28 | M |
| Lacson et al. 2002[13] | 29 | F | Neonatal (died in neonatal period) | NM | Unknown | Congenital transverse fractures of the midshaft of both femora, left radius and ulna | Unknown | No |  |
| Laforgia et al. 2018[14] | 30 | M | Neonatal (died at 1 month) | RYR1-RM | Two variants in the RYR1 gene in exon 28 (g.38964345del (p.Gly1365Glufs∗33) and in exon 104 (g.39076790C>G (p.Phe4976Leu)) | Congenital, displaced fracture of the middle third of the proximal femoral shaft with stumps straddle and a congenital, compound fracture of the middle third of the diaphysis of the left humerus | Unknown | No |  |
| Lal et al. 2016[15] | 31 | M | 17 years (current age) | SEPN1-RM & HRR | homozygous p.G239R mutation in SEPN1 (c.715G>A, NM\_206926.1, p.G239R) | Fracture of the femur (paediatric age) | Unknown | Yes | Serum calcium 2.6; urine calcium/creati-nin 0.59; vitamin D 111 |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Michael et al. 2019[16] | 32 | F | Neonatal;  8 years (current age) | NM | homozygous one-base duplication (c.882dupA, p.Asp295Argfs\*2) in LMOD3 gene | No skeletal fractures | Osteope-nia | No | Daily supplementa-tion with vitamin D |
| Ravenscroft et al. 2013[17] | 33-42 | 10 cases (m = 5, F = 5) | Unknown (mostly neonatal) | NM (KLHL40) | homozygous or compound-heterozygous mutations | Congenital pathological fracture(s) | Unknown | No |  |
| Ryan et al. 2001[18] | 43 - 49 | 7 cases | Neonatal | Severe congenital NM | Unknown | Congenital fractures | 3 patients had generali-zed hypomi-neraliza-tion on skeletal survey | Unknown (probably no) |  |
| 50 | 1 case | Onset in infancy or early childhood | Typical congenital NM | Unknown | Multiple congenital fractures | Unknown | Unknown (probably no) |  |
| 51 - 63 | 13 cases | Onset in childhood | NM | Unknown | Fractures later in life, often after minor trauma | Unknown | Unknown |  |
| Sanchez et al. 2019[19] | 64 | F | Neonatal (died at 8 months) | TTN-CM | Two extremely rare mutations in the titin gene, only expressed in the fetal skeletal transcript of TTN | Multiple congenital long bone fractures | Unknown | No |  |
| 65 | F | Neonatal (alive) | Unknown | Unknown |  |
| Santi et al. 2013[20] | 66-76 | 11 cases | Neonatal | RYR1-CM | Genetically confirmed. Four patients had dominant  RYR1 mutations, and seven had recessive RYR1 mutations. | Congenital fractures of the femur (unknown how many cases) | Unknown | No |  |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Van de Locht et al. 2019[21] | 77 | Unknown | 27 years | NM | mutation in TNNC2 (p.Asp34Tyr) | Unknown | Osteope-nia | Unknown |  |
| Waisayarat et al. 2015[22] | 78 | M | Neonatal (died at 6 weeks) | NM | Heterozygous mutation in ACTA1 in exon 7 (c.1127G > C; Cys376Ser) | Congenital fractures of both humeri and femurs | Unknown | No |  |
| Rodriguez et al. 1988[23] | 79 | M | Neonatal (died at day 1) | Primary myopathic process | Unknown | Congenital Metaphysial detachment of right and left femur and tibia; congenital growth plate fracture of tibia, fibula and ribs | Unknown | No |  |
| 80 | M | Neonatal (died at day 1) | Primary myopathic process | Unknown | Congenital diaphyseal fractur of left and right femur; congenital metaphysial detach of right tibia; congenital growth plate fracture of tibia | Unknown | No |  |
| Han et al. 2013[24] | 81 | M | Neonatal (died at 7 months) | MTM | Mutation in MTM1 gene (c.1261-1C>A in intron 10) | Congenital fracture of both humeri | Unknown | No | Fractures were splinted for treatment within one month |
| Mah et al. 2016[25] | 82 | F | 12 years | RYR1-RM | two heterozygous mutations of the RYR1 gene: missense variant of unknown significance in exon 41; a second gene change in exon 103 | Fracture to the right lateral femoral condyle during a basketball game | Unknown | Yes |  |
| 83 | M | 4 years | CCD | De novo dominant RYR1 mutation involving exon 91 | Fragility fracture of the left humerus (unknown at what age) | Generali-zed osteope-nia | No | Intermittent IV diphospho-nate infusions |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Ilkovski et al. 2004[26] | 84 | F | Neonatal (died 2h after birth) | NM (ACTA1) | heterozygous for a de novo ACTA1 E72K substitution (GAG --> AAG) | Multiple congenital bone fractures | Unknown | No |  |
| Vendittelli et al. 1996[27] | 85 | F | Neonatal (died at 68 days) | NM | Unknown | Congenital diaphyseal fractures of the left femur and humerus | Thin ribs; radiographs of the other long bones appeared normal | No |  |
| Baydan et al. 2018[28] | 86 | M | Neonatal (currently 9 years) | NEM2 | Nonsense mutation in NEB gene (c.10882 A > C, c.4030G > A, c.21627 T > C and c.8499G > A) | Congenital bilateral fracture of femur | Unknown | Yes (at 3 years) |  |
| Lee et al. 2017[29] | 87 | M | 34 years | NEB-related myopathy | Compound heterozygous mutations within the NEB gene (c.9775C.T (p.Arg3259\*) and a splice donor site variant at the 39 end of exon 144 c.21522+3A.G) | Several fractures (unclear at what age) | Unknown | Yes |  |
| Rao et al. 2014[30] | 88 | M | 15 years | Congenital core myopathy | Unknown | Femoral fracture at age 9 years | Unknown | Partially wheelchair bound following femoral fracture |  |
| **Author and year** | **#** | **Gender** | **Age** | **Diagnosis** | **Genetic diagnosis** | **(Congenital) LBFs** | **BMD** | **Ambulant (Yes/No)** | **Remaining** |
| Summers et al. 2013[31] | 89 | F | 16 years | CNM | Unknown | Left subcapital proximal fracture of femur without history of trauma | Osteopo-rosis | No | Long term calcium and vitamin D supplementa-tion |
| Agrawal et al. 2004[32] | 90 | M | Neonatal (died at 6 days) | NM (ACTA1) | c.966AG>G, Asp286Gly | Congenital fracture of femur | Unknown | No |  |
| Wallefeld et al. 2006[33] | 91 | M | Neonatal (currently 1 year) | NM (ACTA1) | c.1248G > T, p.X376TyrextX\*47 | Congenital dislocated fracture of the right humerus | Unknown | No |  |
| Dofash et al. 2021[34] | 92 | M | Neonatal (currently 26 years) | NEM8 | Likely pathogenic variant (c.\*152G>T) in the 3’ untranryr1slated region (UTR) of KLHL40 | Congenital bilateral fractures of femur | Unknown | No (> age 15 years) |  |
| Catterucia et al. 2019[35] | 93 | M | Neonatal (currently 7 months) | RYR1-RM | c.6661A > T/p.(Lys2221\*) and c.13742A > G/p.(Tyr4581Cys) | Multiple congenital fractures of femur, tibia and humerus | Unknown | No |  |

***Abbreviations.*** *LBF = long bone fracture; NM = nemaline myopathy; CCD = central core disease; CNM = centronuclear myopathy; HRR = hereditary hypophosphatemic rickets; NEM2 = nemaline myopathy type 2; NEM8 = nemaline myopathy type 8; LMOD3 = Leiomodin 3; ACTA1 = Actin Alpha 1; KLHL40 = Kelch Like Family Member 40; TNNC2 = Troponin C2; NEB = Nebulin; RYR1(-RM) = Ryanodine receptor type 1(-related myopathy); SELENON(-RM) = SELENON(-related myopathies); (XL-)MTM = (X-linked) myotubular myopathy; ASCC1(-RM) = Activating Signal Cointegrator 1 Complex Subunit 1 (– related myopathy); VMA21 = Vacuolar ATPase Assembly Factor; TTN = titin; TNNC2 = Troponin C2; IV = intravenous.*

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