Supplementary Table 1 Primer sequences of real-time PCR

Gene Name	Forward primer (5'–3')	Reverse primer (5'–3')	Accession number	PCR product size (bp)
actin	GGTATTGTGATGGACTCTGGTGA	TCGGCTGTGGTGGTGAAG	NM_131031	167
clcn7	TCATATAGGCAGGCTGAGCAC	GTCCTGCAAGGTTCTCCACA	NM_001077537	296
sp7	TCCGTATCCATCCGTAGGCT	TTTACCGTACACCTTCCCGC	NM_212863	298
col10a1	CTCCGTGAAAAGCCATGTTGT	TTGCCTGTTGCAGAAATGCC	NM_001083827	324
cx43	TGGCGCATGTCTTCTACCTG	AGAGAGGCTGAAGCCGTAGA	NM_131038	269
dlx2b	CCGCTGTTCTGGATAGCCTT	GCTGGCTCCATACGTTCCAT	NM_131297	290
eve1	TTCACGAGGGAACAGCTGAC	TTCGGGTCCAGTGGATTTGG	NM_131114	194
ctsk	TAGATGCCATGCAGTCCACC	TTCGGTTACGAGCCATCAGG	NM_001017778	201
bmp2b	CAAGCAAATCGGCAGTGGTC	TCTCCGAGAACTTGGTCCCT	NM_131360	292
bmpr1aa	GCCCACCGTATCCAACAGAT	GCACCAGGTCTGATCCAACA	NM_131621	280
bmpr1ab	ATTTTGGAACGCCCACTTGC	ATAAAGGCCTCAATCGGCCC	NM_001004585	153
bmpr1ba	GCGCTACAGTCTCGGTCTAC	GGCCTCCTCTGTGGTGAAAA	NM_131457	247
bmpr1bb	GCAGGACTGATGGTTACTGCT	ACCAATCCTGCGGTTTGTAAC	NM_001114924	239
bmpr2a	ACAACAACAGTCGTCACGGA	TGTCTCCAGAGCTTTTGACCT	NM_001039817	200
bmpr2b	AAATCGTCTGGTACGGCCTG	TCAAAGGACGGGTGGTTTCC	NM_001039807	261
tgfb1a	GTCCGAGATGAAGCGCAGTA	TCAAATGAGAGCCAGCGGTT	NM_182873	189
tgfb1b	TGTGACGTCCACCCTGAAAG	CGTTTTCTGCGCGATTTTGC	XM_687246	240
tgfbr1a	TGAACAGGTACACGGTCACG	GCACCTCAGGGGCCATATAC	NM_001037683	276
tgfbr1b	GAGTCGGCACCAAACGGTAT	CTGCCATCTGTTGGGGATGT	NM_001115059	263
tgfbr2a	ATGTCTGCTCTTCTCCGCAC	AGCATCGGCTTTAGTGGGTC	XM_009293931	209
tgfbr2b	TTGTCTCTGTGACTTCGGGC	CCGATGGCGTTACATCTGGA	NM_182855	216
tgfbr3	ATGCAGGTGTCAAAGACGGT	TCCATAGGCAGGAAGGGGAA	NM_001311172	284
tgfbrap1	CAGAAGCACAGTCAAGCCCT	AGTGTCGCTCGCTCAAGTAG	NM_001083554	269
Gapdh	CATGTTCCAGTATGACTCCACTC	GGCCTCACCCCATTTGATGT	NM_001289726	136
Clcn7	GACAACAGCGAGAATCAGCTC	CCAATGAGGGCACAGATAACC	NM_011930.3	104
Tgfb1	CCGCAACAACGCCATCTATG	CTCTGCACGGGACAGCAAT	NM_011577.2	118
Tgfbr1	GCTGACATCTATGCAATGGG	TTTCTTCAACCGATGGATCA	NM_001312869.1	127
Tgfbr2	CCGCTGCATATCGTCCTGTG	AGTGGATGGATGGTCCTATTACA	NM_009371.3	131
Tgfbr3	CCCTGCATCTGAACCCCATT	CACAGAACCCTCCGAAACCA	NM_011578.4	152
Bmp2	CCGCTGTCTTCTAGTGTTGCT	TCTCTGCTTCAGGCCAAACAT	NM_007553.3	181
Bmpr1a	GATGGTTCGGCAGGTTGGTA	ACGCATTAACACCGTCTGGT	NM_009758.4	148
Bmpr1b	CTCCCTCTGCTGGTCCAAAG	GCTTCCTCCGTGGTGAAGAA	NM_001277217.2	143
Bmpr2	TGGTGAGGATACCAGGCTGA	ACCAAACGATCCAGAACCCC	NM_007561.4	108
Ctsk	TACCCATATGTGGGCCAGGA	TTCAGGGCTTTCTCGTTCCC	NM_007802.4	107
Smad1	GCTTCGTGAAGGGTTGGGG	CGGATGAAATAGGATTGTGGGG	NM_008539.4	147
Smad2	AAGCCATCACCACTCAGAATTG	CACTGATCTACCGTATTTGCTGT	NM_001252481.1	100

Supplementary Table 2 Primer sequences of WISH probes

Gene name	Forward primer (5'–3')	Reverse primer (5'–3')	Accession number	PCR product size (bp)
clcn7	AACAGACAGTTATCCGGGGG	TATCACTGTCAAAGCGGCCA	NM_001077537	864
sp7	GACCCTCACTGGACTGCTTC	CGGCATTTGAGGATTGAGCG	NM_212863	633
col10a1	GCAGTACCAGCCTTACTCCG	AGGTAAACCAACTCCAGGCG	NM_001083827	535
cx43	TGAGGTGGTCTTCCTGGTCA	TCAGTCCTCTAGCGTTGGGA	NM_131038	682
dlx2b	AGCACCGTTCACAAGTCACA	TGCTCTGAAACTGTGTCCCC	NM_131297	617
eve1	CATAAACGAACTTGGCTTGGA	TGTCTGGACGGGGGAATAG	NM_131114	701

Supplementary References

1. Al-Aama JY, Dabbagh AA, Edrees AY. A newly described mutation of the CLCN7 gene causes neuropathic autosomal recessive osteopetrosis in an Arab family. Clin Dysmorphol. 2012;21:1-7.
2. Albuquerque MA, Melo ES, Jorge WA, Cavalcanti MG. Osteomyelitis of the mandible associated with autosomal dominant osteopetrosis: a case report. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2006;102:94-8.
3. Balan A, Girija KL, Ranimol P. Osteomyelitis of maxilla in infantile osteopetrosis: a case report with review of literature. Int J Clin Pediatr Dent. 2011;4:125-8.
4. Batra P, Shah N. Recalcitrant osteomyelitis following tooth extraction in a case of malignant osteopetrosis. Int Dent J. 2004;54:418-23.
5. Besbas N, Draaken M, Ludwig M, Deren O, Orhan D, Bilginer Y, et al. A novel CLCN7 mutation resulting in a most severe form of autosomal recessive osteopetrosis. Eur J Pediatr. 2009;168:1449-54.
6. Celakil T, Dogan M, Rohlig BG, Evlioglu G, Keskin H. Oral rehabilitation of an osteopetrosis patient with osteomyelitis. Case Rep Dent. 2016;2016:6930567.
7. Curran AE, Pfeffle RC, Miller E. Autosomal dominant osteosclerosis: report of a kindred. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 1999;87:600-4.
8. Elster AD, Theros EG, Key LL, Chen MY. Cranial imaging in autosomal recessive osteopetrosis. Part I. Facial bones and calvarium. Radiology. 1992;183:129-35.
9. Eppley BL, Coleman JR. Free fibular flap reconstruction in mandibular osteopetrosis. J Craniofac Surg. 2001;12:369-72.

10. Francisco JV, Reichman L. Osteopetrosis with a complicating osteomyelitis of the mandible. Report of a case. Oral Surg Oral Med Oral Pathol. 1965;19:462-5.
11. Frattini A, Pangrazio A, Susani L, Sobacchi C, Mirolo M, Abinun M, et al. Chloride channel ClCN7 mutations are responsible for severe recessive, dominant, and intermediate osteopetrosis. J Bone Miner Res. 2003;18:1740-7.
12. Gillani S, Abbas Z. Malignant infantile osteopetrosis. J Ayub Med Coll Abbottabad. 2017;29:350-2.
13. Gomes MF, Rangel DC, Starling CC, Goulart M. Familial malignant osteopetrosis in children: a case report. Spec Care Dentist. 2006;26:106-10.
14. Jayachandran S, Kumar MS. A paradoxical presentation of rickets and secondary osteomyelitis of the jaw in Type II autosomal dominant osteopetrosis: Rare case reports. Indian J Dent Res. 2016;27:667-71.
15. Junquera L, Rodriguez-Recio C, Villarreal P, Garcia-Consuegra L. Autosomal dominant osteopetrosis and maxillomandibular osteomyelitis. Am J Otolaryngol. 2005;26:275-8.
16. Kantaputra PN, Thawanaphong S, Issarangporn W, Klangsinsirikul P, Ohazama A, Sharpe P, et al. Long-term survival in infantile malignant autosomal recessive osteopetrosis secondary to homozygous p.Arg526Gln mutation in CLCN7. Am J Med Genet A. 2012;158A:909-16.
17. Krithika C, Neelakandan RS, Sivapathasundaram B, Koteeswaran D, Rajaram PC, Shetkar GS. Osteopetrosis-associated osteomyelitis of the jaws: a report of 4 cases. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2009;108:e56-65.
18. Kulyapina A, Verdaguer MJ, Navarro CC, Navarro VC. Long-term follow-up of bimaxillary osteomyelitis associated with autosomal dominant osteopetrosis: a case report. J Maxillofac

Oral Surg. 2016;15:121-6.

19. Lawoyin DO, Daramola JO, Ajagbe HA, Nyako EA, Lawoyin JO. Osteomyelitis of the mandible associated with osteopetrosis: report of a case. Br J Oral Maxillofac Surg. 1988;26:330-5.

20. Lin WD, Wang CH, Wu KH, Chou IC, Tsai FJ. Identification and characterization of mutations in the CLCN7 gene in a taiwanese patient with infantile malignant osteopetrosis. Pediatr Neonatol. 2016;57:155-7.

21. Liu YP, Lin XH, Yan MY, Lin BQ, Zhuo MY. Debridement in chronic osteomyelitis with benign osteopetrosis: A case report. Exp Ther Med. 2016;12:2811-4.

22. Mainous EG, Hart GB, Soffa DJ, Graham GA. Hyperbaric oxygen treatment of mandibular osteomyelitis in osteopetrosis. J Oral Surg. 1975;33:288-91.

23. Nitzan DW, Marmary Y. Osteomyelitis of the mandible in a patient with osteopetrosis. J Oral Maxillofac Surg. 1982;40:377-80.

24. Ogutcen-Toller M, Tek M, Sener I, Bereket C, Inal S, Ozden B. Intractable bimaxillary osteomyelitis in osteopetrosis: review of the literature and current therapy. J Oral Maxillofac Surg. 2010;68:167-75.

25. Pang Q, Chi Y, Zhao Z, Xing X, Li M, Wang O, et al. Novel mutations of CLCN7 cause autosomal dominant osteopetrosis type II (ADO-II) and intermediate autosomal recessive osteopetrosis (IARO) in Chinese patients. Osteoporos Int. 2016;27:1047-55.

26. Phadke SR, Fischer B, Gupta N, Ranganath P, Kabra M, Kornak U. Novel mutations in Indian patients with autosomal recessive infantile malignant osteopetrosis. Indian J Med Res. 2010;131:508-14.

27. Rashid BM, Rashid NG, Schulz A, Lahr G, Nore BF. A novel missense mutation in the CLCN7 gene linked to benign autosomal dominant osteopetrosis: a case series. J Med Case Rep. 2013;7:7.

28. Rehani U, Adlakha VK, Chandna P, Agarwal A, Rana V, Malik P. Prosthetic rehabilitation in marble bone disease. Int J Clin Pediatr Dent. 2010;3:207-10.

29. Saigal A, Gopal M, Mohanty N, Misra SR. Recurrent osteomyelitis of the mandible in osteopetrosis: a common complication of an uncommon disease. BMJ Case Rep. 2015; 10.1136/2014208974.

30. Satomura K, Kon M, Tokuyama R, Tomonari M, Takechi M, Yuasa T, et al. Osteopetrosis complicated by osteomyelitis of the mandible: a case report including characterization of the osteopetrotic bone. Int J Oral Maxillofac Surg. 2007;36:86-93.

31. Sharma SS, Saravanan C, Sathyabama V, Satish C. Osteopetrosis of the mandible masquerading as tubercular osteomyelitis. BMJ Case Rep. 2013;10.1136/2012007487.

32. Steiner M, Gould AR, Means WR. Osteomyelitis of the mandible associated with osteopetrosis. J Oral Maxillofac Surg. 1983;41:395-405.

33. Sun HJ, Xue L, Wu CB, Zhou Q. Clinical characteristics and treatment of osteopetrosis complicated by osteomyelitis of the mandible. J Craniofac Surg. 2016;27:e728-30.

34. Trivellato AE, Ribeiro MC, Sverzut CE, Bonucci E, Nanci A, de Oliveira PT. Osteopetrosis complicated by osteomyelitis of the maxilla and mandible: light and electron microscopic findings. Head Neck Pathol. 2009;3:320-6.

35. Vazquez E, Lopez-Arcas JM, Navarro I, Pingarron L, Cebrian JL. Maxillomandibular osteomyelitis in osteopetrosis. Report of a case and review of the literature. Oral Maxillofac

Surg. 2009;13:105-8.

36. Yamada T, Mishima K, Imura H, Ueno T, Matsumura T, Moritani N, et al. Osteomyelitis of the mandible secondary to infantile osteopetrosis: a case report. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2009;107:e25-9.

37. Younai F, Eisenbud L, Sciubba JJ. Osteopetrosis: a case report including gross and microscopic findings in the mandible at autopsy. Oral Surg Oral Med Oral Pathol. 1988;65:214-21.

38. Yu T, Yu Y, Wang J, Yin L, Zhou Y, Ying D, et al. Identification of TCIRG1 and CLCN7 gene mutations in a patient with autosomal recessive osteopetrosis. Mol Med Rep. 2014;9:1191-6.

39. Zeng B, Li R, Hu Y, Hu B, Zhao Q, Liu H, et al. A novel mutation and a known mutation in the CLCN7 gene associated with relatively stable infantile malignant osteopetrosis in a Chinese patient. Gene. 2016;576:176-81.

40. Zhang ZL, He JW, Zhang H, Hu WW, Fu WZ, Gu JM, et al. Identification of the CLCN7 gene mutations in two Chinese families with autosomal dominant osteopetrosis (type II). J Bone Miner Metab. 2009;27:444-51.

41. Lu SY, Huang SH, Chen YH. Numb chin with mandibular pain or masticatory weakness as indicator for systemic malignancy–A case series study. J Formos Med Assoc. 2017;116:897-906.