

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- Maroteaux P, Spranger J, Wiedemann HR. Metatrophic dwarfism. *Arch Kinderheilkd* 1966; **173**:211–26.
- Kannu P, Aftimos S, Mayne V, Donnan L, Savarirayan R. Metatropic dysplasia: clinical and radiographic findings in 11 patients demonstrating long-term natural history. *Am J Med Genet A* 2007; **143A**:2512–22.
- Beck M, Roubicek M, Rogers JG, Naumoff P, Spranger J. Heterogeneity of metatropic dysplasia. *Eur J Pediatr* 1983; **140**:231–7.
- Kozlowski K, Beemer FA, Bens G, Dijkstra PF, Iannaccone G, Emons D, Lopez-Ruiz P, Masel J, van Nieuwenhuizen O, Rodriguez-Barrionuevo C. Spondylo-metaphyseal dysplasia (report of 7 cases and essay of classification). *Prog Clin Biol Res* 1982; **104**:89–101.
- Maroteaux P, Spranger J. The spondylometaphyseal dysplasias. A tentative classification. *Pediatr Radiol* 1991; **21**:293–7.
- Shohat M, Lachman R, Gruber HE, Rimoin DL. Brachyolmia: radiographic and genetic evidence of heterogeneity. *Am J Med Genet* 1989; **33**:209–19.
- Vriens J, Watanabe H, Janssens A, Droogmans G, Voets T, Nilius B. Cell swelling, heat, and chemical agonists use distinct pathways for the activation of the cation channel TRPV4. *Proc Natl Acad Sci USA* 2004; **101**:396–401.
- Everaerts W, Nilius B, Owsianik G. The vallinoid transient receptor potential channel Trpv4: From structure to disease. *Prog Biophys Mol Biol* 2009. [Epub ahead of print].
- Muramatsu S, Wakabayashi M, Ohno T, Amano K, Ooishi R, Sugahara T, Shiojiri S, Tashiro K, Suzuki Y, Nishimura R, Kuhara S, Sugano S, Yoneda T, Matsuda A. Functional gene screening system identified TRPV4 as a regulator of chondrogenic differentiation. *J Biol Chem* 2007; **282**:32158–67.
- Masuyama R, Vriens J, Voets T, Karashima Y, Owsianik G, Vennekens R, Lieben L, Torrekens S, Moermans K, Vanden Bosch A, Bouillon R, Nilius B, Carmeliet G. TRPV4-mediated calcium influx regulates terminal differentiation of osteoclasts. *Cell Metab* 2008; **8**:257–65.
- Rock MJ, Prenen J, Funari VA, Funari TL, Merriman B, Nelson SF, Lachman RS, Wilcox WR, Reyno S, Quadrelli R, Vaglio A, Owsianik G, Janssens A, Voets T, Ikegawa S, Nagai T, Rimoin DL, Nilius B, Cohn DH. Gain-of-function mutations in TRPV4 cause autosomal dominant brachyolmia. *Nat Genet* 2008; **40**:999–1003.
- Krakow D, Vriens J, Camacho N, Luong P, Deixler H, Funari TL, Bacino CA, Irons MB, Holm IA, Sadler L, Okenfuss EB, Janssens A, Voets T, Rimoin DL, Lachman RS, Nilius B, Cohn DH. Mutations in the gene encoding the calcium-permeable ion channel TRPV4 produce spondylometaphyseal dysplasia, Kozlowski type and metatropic dysplasia. *Am J Hum Genet* 2009; **84**:307–15.
- Phelps CB, Huang RJ, Lishko PV, Wang RR, Gaudet R. Structural analyses of the ankyrin repeat domain of TRPV6 and related TRPV ion channels. *Biochemistry* 2008; **47**:2476–84.
- Geneviève D, Le Merrer M, Feingold J, Munnich A, Maroteaux P, Cormier-Daire V. Revisiting metatropic dysplasia: presentation of a series of 19 novel patients and review of the literature. *Am J Med Genet A* 2008; **146A**:992–6.

