FOP Learning Center Suggested Readings

General Information

Baujat G, Choquet R, Bouée S, et al. Prevalence of fibrodysplasia ossificans progressiva (FOP) in France: an estimate based on a record linkage of two national databases. *Orphanet J Rare Dis.* 2017;12(1):123. doi:10.1186/s13023-017-0674-5

Connor JM, Evans DA. Fibrodysplasia ossificans progressiva. The clinical features and natural history of 34 patients. *J Bone Joint Surg Br.* 1982;64(1):76-83. doi:10.1302/0301-620X.64B1.7068725

Cohen RB, Burstein AH. Measurements in the conduct of research. *J Bone Joint Surg Am.* 1993;75(3):319-320. doi:10.2106/00004623-199303000-00001

Pignolo RJ, Shore EM, Kaplan FS. Fibrodysplasia ossificans progressiva: diagnosis, management, and therapeutic horizons. *Pediatr Endocrinol Rev.* 2013;10(suppl 2):437-448.

Pignolo RJ, Baujat G, Brown MA, et al. Correction to: Natural history of fibrodysplasia ossificans progressiva: cross-sectional analysis of annotated baseline phenotypes. *Orphanet J Rare Dis.* 2019;14(1):113. doi:10.1186/s13023-019-1096-3

Rogers JG, Geho WB. Fibrodysplasia ossificans progressiva. A survey of forty-two cases. *J Bone Joint Surg Am.* 1979;61(6A):909-914.

Signs and Symptoms

Asadi S, Aranian MR. The role of genetics mutations in genes ACVR1, BMPR1A, BMPR1B, BMPR2, BMP4 in stone man syndrome. *J Hematol Hemother.* 2020;5(1):008.

Kaplan FS, Xu M, Seemann P, et al. Classic and atypical fibrodysplasia ossificans progressiva (FOP) phenotypes are caused by mutations in the bone morphogenetic protein (BMP) type I receptor ACVR1. *Hum Mutat.* 2009;30(3): 379-390. doi:10.1002/humu.20868

Kaplan FS, Zasloff MA, Kitterman JA, et al. Early mortality and cardiorespiratory failure in patients with fibrodysplasia ossificans progressiva. *J Bone Joint Surg Am.* 2010;92(3):686-691. doi:10.2106/JBJS.I.00705

Pignolo RJ, Shore EM, Kaplan FS. Fibrodysplasia ossificans progressiva: clinical and genetic aspects. *Orphanet J Rare Dis.* 2011;6:80. doi:10.1186/1750-1172-6-80

Pignolo RJ, Bedford-Gay C, Liljesthröm M, et al. The natural history of flare-ups in fibrodysplasia ossificans progressiva (FOP): a comprehensive global assessment. *J Bone Miner Res.* 2016;31(3):650-665. doi:10.1002/jbmr.2728

Pignolo RJ, Kaplan FS. Clinical staging of fibrodysplasia ossificans progressiva (FOP). *Bone.* 2018;109:111-114. doi:10.1016/j.bone.2017.09.014

Heterotopic Ossification at Immunization Injection Site

Lanchoney TF, Cohen RB, Rocke DM, Zasloff MA, Kaplan FS. Permanent heterotopic ossification at the injection site after diphtheria-tetanus-pertussis immunizations in children who have fibrodysplasia ossificans progressiva. *J Pediatr.* 1995;126(5 pt 1):762-764. doi:10.1016/s0022-3476(95)70408-6

Spine Anomalies

Schaffer AA, Kaplan FS, Tracy MR, et al. Developmental anomalies of the cervical spine in patients with fibrodysplasia ossificans progressiva are distinctly different from those in patients with Klippel-Feil syndrome: clues from the BMP signaling pathway. *Spine* (Phila Pa 1976). 2005;30(12): 1379-1385. doi:10.1097/01.brs.0000166619.22832.2c

Genetic Testing

Fukuda T, Kanomata K, Nojima J, et al. A unique mutation of ALK2, G356D, found in a patient with fibrodysplasia ossificans progressiva is a moderately activated BMP type I receptor. *Biochem Biophys Res Commun.* 2008;377(3): 905-909. doi:10.1016/j.bbrc.2008.10.093

Gregson CL, Hollingworth P, Williams M, et al. A novel ACVR1 mutation in the glycine/serine-rich domain found in the most benign case of a fibrodysplasia ossificans progressiva variant reported to date. *Bone.* 2011;48(3):654-658. doi:10.1016/j.bone.2010.10.164

Gucev Z, Tasic V, Plaseska-Karanfilska D, et al. Severe digital malformations in a rare variant of fibrodysplasia ossificans progressiva. *Am J Med Genet A.* 2019;179(7):1310-1314. doi:10.1002/ajmg.a.61153

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Kaplan FS, Kobori JA, Orellana C, et al. Multi-system involvement in a severe variant of fibrodysplasia ossificans progressiva (ACVR1 c.772G>A; R258G): a report of two patients. *Am J Med Genet A.* 2015;167A(10):2265-2271. doi:10.1002/ajmg.a.37205

Nakahara Y, Katagiri T, Ogata N, Haga N. ACVR1 (587T>C) mutation in a variant form of fibrodysplasia ossificans progressiva: second report. *Am J Med Genet A.* 2014;164A(1):220-224. doi:10.1002/ajmg.a.36219

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Ohte S, Shin M, Sasanuma H, et al. A novel mutation of ALK2, L196P, found in the most benign case of fibrodysplasia ossificans progressiva activates BMP-specific intracellular signaling equivalent to a typical mutation, R206H. *Biochem Biophy Res Commun.* 2011;407(1):213-218. doi:10.1016/j.bbrc.2011.03.001

Shore EM, Xu M, Feldman GJ, et al. A recurrent mutation in the BMP type I receptor ACVR1 causes inherited and sporadic fibrodysplasia ossificans progressiva. *Nat Genet.* 2006;38(5):525-527. doi:10.1038/ng1783

Whyte MP et al. Fibrodysplasia ossificans progressiva: middle-age onset of heterotopic ossification from a unique missense mutation (c.974G>C, p.G325A) in ACVR1. *J Bone Miner Res.* 2012;27(3):729-737. doi:10.1002/jbmr.1473

Injury and Inflammation

Convente MR, Chakkalakal SA, Yang E, et al. Depletion of mast cells and macrophages impairs heterotopic ossification in an Acvr1 R206H mouse model of fibrodysplasia ossificans progressiva. *J Bone Miner Res.* 2018;33(2):269-282. doi:10.1002/jbmr.3304

Convente MR, Wang H, Pignolo RJ, Kaplan FS, Shore EM. The immunological contribution to heterotopic ossification disorders. *Curr Osteoporos Rep.* 2015;13(2):116-124. doi:10.1007/s11914-015-0258-z

Misdiagnosis/Delayed Diagnosis

Kitterman JA, Kantanie S, Rocke DM, Kaplan FS. Iatrogenic harm caused by diagnostic errors in fibrodysplasia ossificans progressiva. *Pediatrics.* 2005;116(5):e654-e661. doi:10.1542/peds.2005-0469

Clinical Management and Treatment Guidelines

International Clinical Council on FOP (ICC) and Consultants. The medical management of fibrodysplasia ossificans progressiva: current treatment considerations. Revised January 2020. Accessed December 8, 2020. http://www.iccfop.org/dvlp/wp-content/uploads/2020/03/Guidelines_January-2020.pdf

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