

# **INGEMM - Institute of Medical and Molecular Genetics**

## **PUBLICATIONS**

- \* Barraza-García J, Rivera-Pedroza CI, Belinchón A, Fernández-Camblor C, Valenciano-Fuente B, Lapunzina P, Heath KE. A novel SMARCAL1 missense mutation that affects splicing in a severely affected Schimke immunoosseous dysplasia patient. *Eur J Med Genet.* 2016;59(8):363-6. Article. IF: 2.137; Q3
- \* Barraza-García J, Rivera-Pedroza CI, Salamanca L, Belinchón A, López-González V, Senthordi-Montane L, del Pozo A, Santos-Simarro F, Campos-Barros A, Lapunzina P, Guillén-Navarro E, González-Casado I, García-Miñaur S, Heath KE. Two Novel POC1A Mutations in the primordial Dwarfism, SOFT syndrome: clinical homogeneity but also unreported malformations. *Am J Med Genet A.* 2016;170(1):210-6. Article. IF: 2.259; Q3
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- \* Fergelot P, Van Belzen M, Van Gils J, Afenjar A, Armour CM, Arveiler B, Beets L, Burglen L, Busa T, Collet M, Deforges J, de Vries BBA, Garrido ED, Dorison N, Dupont J, Francannet C, García-Miñaur S, Vila EG, Gebre-Medhin S, Querol BG, Genevieve D, Gerard M, Gervasini CG, Goldenberg A, Josifova D, Lachlan K, Maas S, Maranda B, Moilanen JS, Nordgren A, Parent P, Rankin J, Reardon W, Rio M, Roume J, Shaw A, Smigiel R, Sojo A, Solomon B, Stembalska A, Stumpel C, Suarez F, Terhal P, Thomas S, Touraine R, Verloes A, Vincent-Delorme C, Wincent J, Peters DJM, Bartsch O, Larizza L, Lacombe D, Hennekam RC. Phenotype and genotype in 52 patients with rubinstein-taybi syndrome caused by EP300 mutations. *Am J Med Genet A.* 2016;170(12):3069-82. Article. IF: 2.259; Q3
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- \* Haghghi A, Kavehmanesh Z, Haghghi A, Salehzadeh F, Santos-Simarro F, Van Maldergem L, Cimbalistiene L, Collins F, Chopra M, Al-Sinani S, Dastmalchian S, de Silva DC, Bakhti H, Garg A, Hilbert P. Congenital generalized lipodystrophy: identification of novel variants and expansion of clinical spectrum. *Clin Genet.* 2016;89(4):434-41. Article. IF: 3.326; Q2

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