

Dr Ana Beleza's publications

- Elouej S, Beleza-Meireles A, Caswell R, Colclough K, Ellard S, Desvignes JP, B Christophee, Levy N, Mohammed S, Desandre-Giovannali A. Exome sequencing reveals a de novo POLD1 mutation causing phenotypic variability in mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL). Manuscript Number: METABOLISM-D-16-00840R2 (in press).
- Loureiro S , Almeida J, Café C , Mouga S, Beleza A, Oliveira B, Sá J, Carreira I, Saraiva J , Vicente A, Oliveira G. Copy number variations in chromosome 16p13.11 - the neurodevelopmental clinical spectrum. Manuscript Autism Research (AUR-16-0093) (in press).
- Beleza-Meireles A, Steenhaut P, Hocq C, Clapuyt P, Bernard P, Debauche C, Sznajer Y. "Serpentine-like syndrome"-A very rare multiple malformation syndrome characterised by brachioesophagus and vertebral anomalies. *Eur J Med Genet*. 2016. pii: S1769-7212(16)30178-1.
- Carreira IMM, Ferreira SI, Matoso E, Pires LM, Ferrão J, Jardim AC, Mascarenhas A, Pinto M, Lavoura N, Pais C, Paiva P, Simões L, Caramelo F, Ramos L, Venâncio M, Ramos L, Beleza A, Sá J, Saraiva J, Barbosa-de-Melo J. Copy Number Variants Prioritization after array-CGH analysis - Portuguese experience from 1000 patients. *Mol Cytogenet*. 2015;8:103.
- Yilmaz R, Beleza-Meireles A, Price, S, Oliveira R, Kubisch C, Clayton-Smith J, Szakszon K, Borck G. A Recurrent Synonymous KAT6B Mutation Causes Say-Barber- Biesecker/Young-Simpson Syndrome by Inducing Aberrant Splicing. *American Journal of Human Genetics*, 2015.
- Carreira IMM, Ferreira SI, Matoso E, Pires LM, Ferrão J, Jardim AC, Mascarenhas A, Pinto M, Lavoura N, Pais C, Paiva P, Simões L, Caramelo F, Ramos L, Venâncio M, Ramos L, Beleza A, Sá J, Saraiva J, Barbosa-de-Melo J. Copy Number Variants Prioritization after array-CGH analysis - Portuguese experience from 1000 patients. *Molecular Cytogenetics*, 2015.
- Beleza-Meireles A, Hart R, Clayton-Smith J, Oliveira R, Reis CF, Venâncio M, Ramos F, Sá J, Ramos L, Cunha E, Pires LM, Carreira IM, Scholey R, Wright E, Urquhart JE, Briggs TA, Kerr B, Kingston H, Metcalfe K, Donnai D, Newman WG, Saraiva JM, Tassabehji M. Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. *Eur J Med Genet*, 2015.
- Beleza-Meireles A, Clayton-Smith J, Saraiva JM, Tassabehji M. Oculo-auriculo-vertebral spectrum: a review of the literature and genetic update. *J Med Genet*, 2014.
- Beleza-Meireles A, Matoso E, Ramos L, Melo JB, Carreira IM, Silva ED, Saraiva JM. Cryptic 7q36.2q36.3 deletion causes multiple congenital eye anomalies and craniofacial dysmorphism. *Am J Med Genet A*, 2013.
- Zimoń M, Baets J, Almeida-Souza L, De Vriendt E, Nikodinovic J, Parman Y, Battaloğlu E, Zeliha Matur Z, Guergueltcheva V, Tournev I, Auer-Grumabach M, De Rijk P, Britt-Sabina Petersen BS, Müller T, Fransen E, Van Damme P, Löscher WN, Barišić N, Mitrovic Z, Previtali SC, Topaloğlu H, Bernert G, Beleza-Meireles A, Todorovic S, Savic- Pavicevic D, Ishpekova B,

- Lechner S, Peeters K, Ooms T, Hahn AF, Züchner S, Timmerman V, Van Dijck P, Rasic VM, Janecke AR, De Jonghe P & Jordanova A. Loss of function mutations in HINT1 are a major cause of autosomal recessive axonal neuropathy peripheral neuropathy with neuromyotonia. *Nat Genet*, 2012.
- Beleza-Meireles A, Cerqueira R, Sousa SB, Palmeiro A, Ramos L. Novel deletion encompassing exons 5-12 of the UBE3A gene in a girl with Angelman syndrome. *Eur J Med Genet*. 2011.
 - Simpson CL, Lemmens R, Miskiewicz K, Broom WJ, Hansen VK, van Vught PW, Landers JE, Sapp P, Van Den Bosch L, Knight J, Neale BM, Turner MR, Veldink JH, Ophoff RA, Tripathi VB, Beleza A, Shah MN, Proitsi P, Van Hoecke A, Carmeliet P, Horvitz HR, Leigh PN, Shaw CE, van den Berg LH, Sham PC, Powell JF, Verstreken P, Brown RH Jr, Robberecht W, Al-Chalabi A. Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. *Hum Mol Genet*, 2009.
 - Diekstra FP, Beleza-Meireles A, Leigh NP, Shaw CE, Al-Chalabi A. Interaction between PON1 and population density in amyotrophic lateral sclerosis. *Neuroreport*, 2009.
 - Beleza-Meireles A, Al-Chalabi A. Genetic studies of amyotrophic lateral sclerosis– Controversies and perspectives. *Amyotroph Lateral Scler*, 2008.
 - Beleza-Meireles A, Kockum I, Yuan QP, Wetterberg L, Gustavson KH, Schalling M. Complex aetiology of an apparently autosomal recessive form of unspecific mental retardation. *BMC Medical Genetics*, 2008.
 - Beleza-Meireles A, Töhhönen V, Radmayr C, Schwentner C, Söderhäll C, Kockum I, Nordenskjöld A. Activating transcription factor 3: a hormone responsive gene in the aetiology of hypospadias. *Eur J Endocrinol*, 2008.
 - Beleza-Meireles A, Kockum I, Lundberg F, Söderhäll C, Nordenskjöld A. Risk factors for hypospadias in the estrogen receptor 2 gene. *J Clin Endocrinol Metab*, 2007.