

- in seipin gene in a patient with Berardinelli-Seip congenital lipodystrophy and dystonia: phenotype variability suggests multiple roles of seipin gene[J]. *J Neurol Neurosurg Psychiatry*, 2009, 80(10): 1180-1181.
- [17] Shirwalkar HU, Patel ZM, Magre J, et al. Congenital generalized lipodystrophy in an Indian patient with a novel mutation in BSCL2 gene[J]. *J Inher Metab Dis*, 2008, 31(2): 317-322.
- [18] Ebihara K, Kusakabe T, Masuzaki H, et al. Gene and phenotype analysis of congenital generalized lipodystrophy in Japanese: a novel homozygous nonsense mutation in seipin gene[J]. *J Clin Endocrinol Metab*, 2004, 89(5): 2360-2364.
- [19] Nishiyama A, Yagi M, Awano H, et al. Two Japanese infants with congenital generalized lipodystrophy due to BSCL2 mutations[J]. *Pediatr Int*, 2010, 51(6): 775-779.
- [20] Rahman OU, Khawar N, Khan MA, et al. Deletion mutation in BSCL2 gene underlies congenital generalized lipodystrophy in a Pakistani family[J]. *Diagn Pathol*, 2013, 8: 78.
- [21] Haghighi A, Kavehmanesh Z, Haghighi A, et al. Congenital generalized lipodystrophy: identification of novel variants and expansion of clinical spectrum[J]. *Clin Genet*, 2016, 89(4): 434-441.
- [22] Opri R, Fabrizi GM, Cantalupo G, et al. Progressive myoclonus epilepsy in congenital generalized lipodystrophy type 2: Report of 3 cases and literature review[J]. *Seizure*, 2016, 42: 1-6.
- [23] Purizaca-Rosillo N, Mori T, Benites-Cóndor Y, et al. High incidence of BSCL2 intragenic recombinational mutation in peruvian type 2 Berardinelli-Seip syndrome[J]. *Am J Med Genet A*, 2017, 173(2): 471-478.
- [24] Agarwal AK, Simha V, Oral EA, et al. Phenotypic and genetic heterogeneity in congenital generalized lipodystrophy[J]. *J Clin Endocrinol Metab*, 2003, 88(10): 4840-4847.
- [25] Lundin C, Nordström R, Wagner K, et al. Membrane topology of the human seipin protein[J]. *FEBS Lett*, 2006, 580(9): 2281-2284.
- [26] Sim MF, Talukder MU, Dennis RJ, et al. Analyzing the functions and structure of the human lipodystrophy protein seipin[J]. *Methods Enzymol*, 2014, 537: 161-175.
- [27] Garg A, Misra A. Lipodystrophies: rare disorders causing metabolic syndrome[J]. *Endocrinol Metab Clin North Am*, 2004, 33(2): 305-331.
- [28] Diker-Cohen T, Cochran E, Gorden P, et al. Partial and generalized lipodystrophy: comparison of baseline characteristics and response to metreleptin[J]. *J Clin Endocrinol Metab*, 2015, 100(5): 1802-1810.
- [29] Patni N, Garg A. Congenital generalized lipodystrophies-new insights into metabolic dysfunction[J]. *Nat Rev Endocrinol*, 2015, 11(9): 522-534.

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