Convergence - Oriented Laboratory Medicine

LMCE 2020 ANNUAL & KSLM MEETING

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Main Scientific Publications:

- 1. Shinn Young Kim, Hyun-Sun Ko, Namshin Kim, Seon-Hee Yim, Seung-Hyun Jung, Jiwoong Kim, Myung-Duk Lee, Yeun-Jun Chung. A missense mutation in EBF2 was segregated with imperforate anus in a family across three generations. American Journal of Medical Genetics Part A, 176(7) 1632-1636 (2018). # Co-first author
- 2. Seongmun Jeong, Jiwoong Kim, Won Park, Hongmin Jeon, Namshin Kim. SEXCMD: Development and validation of sex marker sequences for whole-exome/genome and RNA sequencing. PloS one, 12(9) e0184087 (2017). # Corresponding author
- 3. Hyeong-Gon Moon, Namshin Kim, Seongmun Jeong, Minju Lee, HyunHye Moon, Jongjin Kim, Tae-Kyung Yoo, Han-Byoel Lee, Jisun Kim, Dong-Young Noh, Wonshik Han. The Clinical Significance and Molecular Features of the Spatial Tumor Shapes in Breast Cancers. PloS one, 10(12) e0143811 (2015). # Co-first author
- 4. Mi-Ae Jang, Sang-Heon Lee, Namshin Kim, Chang-Seok Ki. Frequency and spectrum of actionable pathogenic secondary findings in 196 Korean exomes. Genetics in Medicine, 17(12) 1007-1011 (2015). # Co-corresponding author
- 5. Hee Jeong Yoo, Kyung Kim, In Hyang Kim, Seong-Hwan Rho, Jong-Eun Park, Ki Young Lee, Soon Kim, Byung Yoon Choi, Namshin Kim. Whole Exome Sequencing for a Patient with Rubinstein-Taybi Syndrome Reveals de Novo Variants besides an Overt CREBBP Mutation. International journal of molecular sciences, 16(3) 5697-5713 (2015). # Corresponding author
- 6. Byung Yoon Choi, Jiwoong Kim, Juyong Chung, Ah Reum Kim, Sue Jean Mun, Seong Il Kang, Sang-Heon Lee, Namshin Kim, Seung-Ha Oh. Whole-exome sequencing identifies a novel genotype-phenotype correlation in the entactin domain of the known deafness gene TECTA. PloS one, 9(5) e97040 (2014). # Co-corresponding author
- 7. Byung-Joo Min, Namshin Kim, Taesu Chung, Ok-Hwa Kim, Gen Nishimura, Chin Youb Chung, Hae Ryong Song, Hyun Woo Kim, Hye Ran Lee, Jiwoong Kim, Tae-Hoon Kang, Myung-Eui Seo, San-Deok Yang, Do-Hwan Kim, Seung-Bok Lee, Jong-Il Kim, Jeong-Sun Seo, Ji-Yeob Choi, Daehee Kang, Dongsup Kim, Woong-Yang Park, Tae-Joon Cho. Whole-Exome Sequencing Identifies Mutations of KIF22 in Spondyloepimetaphyseal Dysplasia with Joint Laxity, Leptodactylic Type. The American Journal of Human Genetics, 89(6) 760-766 (2011). # Co-first author

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