Digital Transformation of Laboratory Medicine Linchpin of Future Medical Value



OCTOBER, 26-28, 2022 Swiss Grand Hotel, Seoul, Korea

## **CURRICULUM VITAE**

Name	Murim Choi	Country	Korea
<b>Current Position</b>	Professor Department of Biomedical Sciences Seoul National University of College of Medicine		

Educational Background			
2006	Ph.D.	Duke University (Genetics and Developmental Biology)	
2000	M.S	Seoul National University (Molecular Biology)	
1998	B.S.	Seoul National University (Molecular Biology)	

Professional Experience		
2007-2013	Postdoc Researcher, Department of Genetics	
	Yale University, New Haven, CT, USA	
2013-present	Professor, Department of Biomedical Sciences	
	Seoul National University of College of Medicine Associate Professor	

## **Professional Organizations**

Korean Society of Molecular and Cellular Biology (KSMCB)

Korea Genome Organization (KOGO)

Korean Society of Biochemistry and Molecular Biology (KSBMB)

## **Main Scientific Publications**

- Hong et el., (2022) Transcriptome-based variant calling and aberrant mRNA discovery enhance diagnostic efficiency for neuromuscular diseases. J Med Genet 2021-1083
- Yoo et al., (2021) Disease-specific eQTL screening reveals an anti-fibrotic effect of AGXT2 in non-alcoholic fatty liver disease. J Hepatol. 75:514
- Lee et al., (2021) Somatic uniparental disomy mitigates the most damaging EFL1 allele combination in Shwachman-Diamond syndrome. Blood 138:2117
- Yoo et al., (2017) GABBR2 mutations determine phenotype in Rett syndrome and epileptic



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encephalopathy. Ann Neurol 82:466.

- Seo et al., (2017) Tofacitinib relieves symptoms of stimulator of interferon genes (STING)associated vasculopathy with onset in infancy caused by 2 de novo variants in TMEM173. J
  Allergy Clin Immunol 139:1396.
- Lee et al., (2015) Abatacept alleviates severe autoimmune symptoms in a patient carrying a de novo variant in CTLA-4. J Allergy Clin Immunol. 137:327