



- **Name:** Chang-Seok Ki

- **Current Position:**

Professor, Department of Laboratory Medicine and Genetics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea

- **Country:** Korea

- **Educational Background:**

2004 Ph.D. in Medical Science, Postgraduate School, Sungkyunkwan University School of Medicine, Seoul, Korea

1998 M.S. in Medical Science, Postgraduate School, Korea University, College of Medicine, Seoul, Korea

1994 B.S. in Medicine, Seoul National University, College of Medicine, Seoul, Korea

- **Professional Experiences:**

2015-present Professor
Department of Laboratory Medicine and Genetics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea

2009-2015 Associate Professor
Department of Laboratory Medicine and Genetics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea

2008-2009 Visiting Scholar
Duke Center for Human Genetics
Duke University Medical Center, Durham, NC, USA

2005-2009 Assistant Professor
Department of Laboratory Medicine and Genetics, Samsung



Medical Center, Sungkyunkwan University School of
Medicine, Seoul, Korea

2003-2005 Clinical Professor

Department of Laboratory Medicine, Samsung Medical
Center, Sungkyunkwan University School of Medicine, Seoul,
Korea

2001-2003 Clinical Fellow

Department of Laboratory Medicine, Samsung Medical
Center, Sungkyunkwan University School of Medicine, Seoul,
Korea

1995-1999 Resident

Department of Laboratory Medicine, Samsung Medical
Center, Sungkyunkwan University School of Medicine, Seoul,
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• Professional Organizations

Department of Laboratory Medicine & Genetics

Samsung Medical Center, Sungkyunkwan University School of Medicine

• Main Scientific Publications:

Selected publications as 1st or corresponding author (Jan 2015 ~ Apr 2016)

1. Huh HJ, Koh WJ, Song DJ, Ki CS, Lee NY. Evaluation of the Cobas TaqMan MTB Test for the Detection of Mycobacterium tuberculosis Complex According to Acid-Fast Bacilli Smear Grades in Respiratory Specimens. *J Clin Microbiol* 2015;53(2):696-698
2. Kim YE, Oh KW, Kwon MJ, Choi WJ, Oh SI, Ki CS, Kim SH. De novo FUS mutations in 2 Korean patients with sporadic amyotrophic lateral sclerosis. *Neurobiol Aging* 2015;36(2):1604.e17-19
3. Huh HJ, Jang MA, Seo JY, Kim JY, Ki CS, Kim JW, Lee NY. Evaluation of the iNtRON VRE vanA/vanB Real-Time PCR Assay for Detection of Vancomycin-Resistant Enterococci. *Ann Lab Med* 2015;35(1):76-81

Somatic KRAS Mutation in a Korean Baby with Nevus Sebaceus Syndrome.

Ann Lab Med 2015;35(1):178-180

5. Huh HJ, Kwon HJ, Ki CS, Lee NY. Comparison of the GENEDIA MTB Detection Kit and the COBAS TaqMan MTB Assay for the Detection of Mycobacterium tuberculosis in Respiratory Specimens. J Clin Microbiol 2015;53(3):1012-1014

6. Jang MA, Kim EK, Now H, Nguyen NT, Kim WJ, Yoo JY, Lee J, Jeong YM, Kim CH, Kim OH, Sohn S, Nam SH, Hong Y, Lee YS, Chang SA, Jang SY, Kim JW, Lee MS, Lim SY, Sung KS, Park KT, Kim BJ, Lee JH, Kim DK, Kee C, Ki CS. Mutations in DDX58, which Encodes RIG-I, Cause Atypical Singleton-Merten Syndrome. Am J Hum Genet. 2015;96(2):266-274

7. Jang MA, Lee SH, Kim N, Ki CS. Frequency and spectrum of actionable pathogenic secondary findings in 196 Korean exomes. Genet Med 2015;17(12):1007-1011

8. Song JS, Lim DH, Chung ES, Chung TY, Ki CS. Mutation Analysis of the TGFBI Gene in a Consecutive Series of Korean Patients with Corneal Dystrophies. Ann Lab Med 2015;35(3):336-340

9. Jang MA, Lee T, Lee J, Cho EH, Ki CS. Identification of a Novel De Novo Variant in the PAX3 Gene in Waardenburg Syndrome by Diagnostic Exome Sequencing: The First Molecular Diagnosis in Korea. Ann Lab Med 2015;35(3):362-365

10. Jang MA, Kim OH, Kim SW, Ki CS. Identification of p.Glu131Lys Mutation in the IHH Gene in a Korean Patient With Brachydactyly Type A1. Ann Lab Med 2015;35(3):387-389