



Posters: Genetic Counseling and Clinical Testing | #1654 | Wednesday 5 - 7 p.m.
Molecular genetic testing of podocin (NPHS2) mutations in steroid resistant nephrotic syndrome by high resolution melting curve analysis.

M. Lu, et al., Dept. of Pathology and Lab Medicine, UNC at Chapel Hill, Chapel Hill, NC

Posters: Cardiovascular Genetics | #753 | Wednesday 5 - 7 p.m.
Is E-selectin Gene S128R Polymorphism Associated with Incidence or Severity of Kasawaki Disease?

S. Toshihiko, et al., Nagasaki University Hospital, nagasaki-shi nagasaki-ken, Japan.

Posters: Mol. Basis of Mendelian Disorders | #2204 | Wednesday 5 - 7 p.m.
Mutation screening and cellular complementation aid gene identification in mitochondrial complex I deficiency.

T.B.L. Haack, et al., HealthInstitute of Human Genetics, Helmholtz Zentrum München, German Research Center for Environmental Health, Neuherberg, Germany

Posters: Genetic Counseling and Clinical Testing | #1641 | Wednesday 5 - 7 p.m.
Mutation Analysis of SLC26A4 for Pendred Syndrome and Non-Syndromic Hearing Loss by High Resolution Melting.

N. Chen, I. Schrijver, Dept Pathology, Stanford Univ, Palo Alto, CA.

Posters: Genetic Counseling and Clinical Testing | #1643 | Wednesday 5 - 7 p.m.
Rapid identification of common -thalassemia mutations in the Chinese population using duplex or triplex amplicon genotyping by high resolution melting analysis.

Z. Ren, et al., Shanghai Inst Med Gen, Shanghai Children's Hosp, Shanghai Jiaotong Univ School of Medicine, 24/1400 West Beijing Rd, Shanghai 200040, China.

Posters: Cancer Genetics | #516 | Thursday 5 - 7 p.m.
Detection of BRCA1 and BRCA2 mutations by High-Resolution Melting curve analysis in breast cancer patients from Thailand and Pakistan.

J. Ahmad, et al., Genetic Cancer Susceptibility, International Agency for Research on Cancer, Lyon, France;

Posters: Genomics | #1890 | Thursday 5 - 7 p.m.
Detecting Mutant Allele Fractions as low as 0.01% by combining Digital PCR, LunaProbes™ and Mutant Allele Amplification Bias (MAAB) Methods.

M.D. Poulson, et al., Idaho Technology, Inc., Salt Lake City, UT.

Posters: Metabolic Disorders | #2009 | Thursday 5 - 7 p.m.
Assessment of a high-throughput DNA melting analysis assay for rapid screening of gene variants in the Ornithine Transcarbamylase Gene.

K. Sumner, et al., ARUP Institute for Clinical and Experimental Pathology, Salt Lake City, UT

Posters: Metabolic Disorders | #2043 | Thursday 5 - 7 p.m.
A Study of Wilson Disease Mutations in Spain.

C. Solis-Villa, et al., Hepatology, University Hospital La Paz, Madrid, Spain

Posters: Cancer Genetics | #469 | Thursday 5 - 7 p.m.
EMSY and prostate cancer.

R. Nurminen, et al., Laboratory of Cancer Genetics, Institute of Medical Technology, University of Tampere and Tampere University Hospital, Tampere, Finland

Posters: Mol. Basis of Mendelian Disorders | #2303 | Fri. 10:30 - 12:30 p.m.
An Analysis of DNA Sequence Variation in the C2ORF71 Gene.

P.I. Sergouniotis^{1,2}, et al., UCL Institute of Ophthalmology, London, EC1V 9EL, United Kingdom.

Posters: Psych. Gen., Neurogen. and Neurodegen. | #2667 | Fri. 10:30 - 12:30 p.m.
Molecular analysis of REEP1 gene mutations in patients with familial and sporadic spastic paraplegia.

D. Di Bella, et al., Genet Neurodegen Metab Dis, IRCCS Ist Neurol Carlo Besta, Milan, Italy

Posters: Psych. Gen., Neurogen. and Neurodegen. | #2680 | Fri. 10:30 - 12:30 p.m.
The development of high-throughput gene scanning system for autism spectrum disorders by a PCR coupled high-resolution melting curve analysis.

K. Yanagi¹, et al., Dept Medical Genetics, Univ Ryukyus, Nishihara, Japan



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Hi-Res Melting Applications

Concurrent Platform Session V (57-64)

Presentation: #357

Time: Friday, Nov. 5, ~5:00 p.m.

SHOC2 mutation analysis in Noonan-like syndrome and hematologic malignancies.

S. Komatsuzaki, Y. Aoki, T. Niihori, N. Okamoto, R.C.M. Hennekam, S. Hopman, H. Ohashi, S. Mizuno, Y. Watanabe, H. Kamasaki, I. Kondo, N. Moriyama¹⁰, K. Kurosawa, H. Kawame, M. Imaizumi¹, T. Rikiishi, S. Tsuchiya, S. Kure, Y. Matsubara. **Department of Medical Genetics, Tohoku University School of Medicine, Sendai, Japan;**

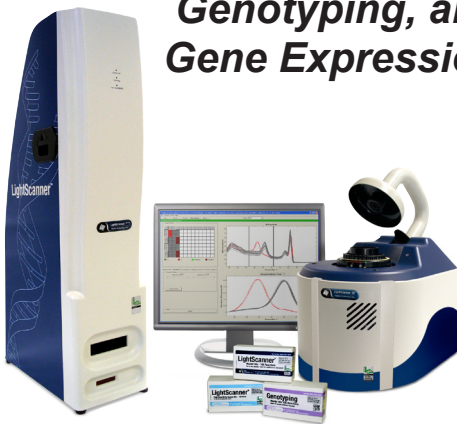
Presentation: #365

Time: Friday, Nov. 5, ~5:15 p.m.

Keratosis Follicularis Spinulosa Decalvans is caused by mutations in MBTPS2

E. Aten, L. Brasz, D. Bornholdt, I.B. Hooijkaas, M.E. Porteous, V.P. Sybert, M.H. Vermeer, R.H.A.M Vossen, M.J.R. van der Wielen¹, E. Bakker, M.H. Breuning, K.H. Grzeschik, J.C. Oosterwijk, J.T. den Dunnen¹. **Center of Human and Clinical Genetics, Leiden University Medical Center, Leiden, the Netherlands;**

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