



nederlandse vereniging voor

Humane Genetica

Najaarssymposium

vrijdag 19 november 2010

AMC, Collegezaal 4, Meibergdreef 9, 1105 AZ Amsterdam

09.00- 10.00	Ontvangst NVHG-leden; koffie / thee
10.00-10.05	Opening
10.05-10.20	Chantal Stoepker - VUMC SLX4, a coordinator of structure-specific endonucleases, is mutated in a new Fanconi anemia subtype and possibly associated with hereditary breast cancer
10.20-10.35	Hans Dauwerse - LUMC Identification of 2 novel genes involved in Treacher Collins syndrome
10.35-10.50	Margit Schraders - UMCN Causes of Autosomal Recessive Nonsyndromic Hearing Impairment in the Netherlands
10.50-11.05	Arjan Buijs - UMCU A new pathogenomic mechanism in FPD/AML by a constitutional t(16;21) involving 16p13 ATF7IP2 and 21q22 RUNX1 loci.
11.05-11.20	Richard Lemmers - LUMC A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy
11.20-11.35	Aida Bertoli-Avella - ErasmusMC Mutations in SMAD3 in the TGF- β Pathway cause a New Syndromic Form of Aortic Aneurysms and Dissections with Early-Onset Osteoarthritis
11.35-11.50	Gajja Salomons - VUMC IDH2 Mutations in Patients with D-2-Hydroxyglutaric Aciduria type II
12:00-12.20	NVHG ledenvergadering
12.20-12.35	VKGL ledenvergadering
12.35-14.00	Lunch en posters
14.00-14.15	Lude Franke - UMCG Connecting genotypes with phenotypes: a map of intermediate genes
14.15-14.30	Mike Gerards - UMCM Riboflavin responsive OXPHOS complex I deficiency caused by defective ACAD9: new function for an old gene
14.30-14.45	Melvin Evers - LUMC Reduction of Prolonged CAG Repeat Containing Alleles in Huntington's Disease Using Antisense Oligonucleotides
14.45-15.00	Kornelia Neveling - UMCN Design and validation of a next generation sequencing approach for hereditary blindness in diagnostics
15.00-15.15	Lisenka Vissers - UMCN A de novo paradigm for mental retardation
15.15-15.30	Pascal Borry - Leuven Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes
15.30-16.00	Theepauze
16.00-16.10	Uitreiking ESHG travel-award
16.10-16.40	Edwin Cuppen - UMCU The next-generation sequencing revolution: applications in genetic research and diagnostics
16.50	Afsluiting / Drankjes en hapjes.