

Programme CANCER GENETICS MEETING

(changes reserved)

Wednesday 10 March 2010

9:00	Registration and coffee	14:10	(12) Identification of multiple genetic variants which predispose to prostate cancer by genome-wide association in the PRACTICAL international consortium – <i>Prof Rosalind Eeles</i>
10:00-10:05	Opening address – <i>Prof Anneke Lucassen and Dr Frederik Hes</i>	14:30	(13) News on BRCAx genes – <i>Prof Peter Devilee</i>
10:05-12:15	Hereditary gastrointestinal cancers Chairs: <i>Dr Eamonn Sheridan and Dr Rolf Sijmons</i>		SUBMITTED ABSTRACTS
10:05	(1) HLA class I defects in DNA-repair deficient colorectal cancers: implications for therapy and prevention of colorectal cancer syndromes – <i>Noel de Miranda</i>	14:50	(14) The IMPACT study: Identification of Men with a genetic predisposition to ProstAte Cancer: Targeted screening in BRCA1/2 mutation carriers and controls. Preliminary results <i>Dr Elena Castro</i>
10:25	(2) An emerging hereditary stomach cancer protocol – <i>Dr Irma Kluijt</i>	15:00	(15) Long term outcome of breast cancer in women aged 30 years or younger, based on family history, pathology and BRCA1/BRCA2/TP53 status – <i>Prof Gareth Evans</i>
10:45	(3) Patients with Lynch syndrome caused by an MSH2 mutation run an increased risk of urinary bladder cancer <i>Prof Nicoline Hoogerbrugge</i>	15:10	(16) Breast cancer susceptibility variants modify risk in familial ovarian cancer – <i>Ayse Latif</i>
11:05	(4) An overview of Lynch syndrome families with an EPCAM 3' end deletion – <i>Dr Marjolijn Ligtenberg</i>	15:20	(17) Unclassified variants in BRCA1 end BRCA2: does in silico analysis discriminate between class II versus class III unclassified variants in breast/ovarian cancer families? – <i>Nandy Hofland</i>
	SUBMITTED ABSTRACTS	15:30-16:00	TEA
11:25	(5) Psychological distress after MSI testing shortly following a diagnosis of colorectal cancer – <i>Karin Landsbergen</i>	16:00-18:00	Endocrine and other cancer syndromes Chairs: <i>Prof Anneke Lucassen and Dr Frederik Hes</i>
11:35	(6) Burden of undergoing pancreatic screening in high-risks individuals; first experiences – <i>Dr Femme Harinck</i>	16:00	(18) SDHAF2 mutations in paraganglioma and pheochromocytoma <i>Dr Jean-Pierre Bayley</i>
11:45	(7) Familial adenomatous polyposis (FAP): attitudes towards genetic testing in childhood and reproductive decision-making <i>Dr Kirsten Douma</i>	16:20	(19) Psychological aspects in Li-Fraumeni syndrome (LFS) and Von Hippel-Lindau disease (VHL) – <i>Chantal Lammens</i>
11:55	(8) A substantial number of patients participating in a population based colorectal cancer screening programme have an increased familial risk – <i>Nicky Dekker</i>	16:40	(20) Retinoblastoma not just a paradigm for Cancer Genetics but a possible window into the future of Medical Genetics <i>Dr Trevor Cole</i>
12:05	(9) Two PTEN mutation carriers with early onset colorectal cancer <i>Dr Rogier Kersseboom</i>		SUBMITTED ABSTRACTS
12:15-13:30	LUNCH and POSTER VIEWING	17:00	(21) Risk of second primary malignancies after hereditary retinoblastoma according to type of RB1-mutation <i>Dr Charlotte Dommering</i>
13:30-15:30	Hereditary Breast Ovarian cancers Chairs: <i>Prof Gareth Evans and Prof Nicoline Hoogerbrugge</i>	17:10	(22) Hereditary leiomyomatosis and renal cell cancer, a nation-wide study of 32 families referred for fumarate hydratase germline mutation analysis – <i>Diederik L. Smit</i>
13:30	(10) Use of hormone replacement therapy in women after bilateral salpingo-oophorectomy (BSO) in BRCA1/2 carriers. <i>Prof Anthony Howell</i>	17:20	(23) Localization effects of three novel mutations in the fumarate hydratase gene – <i>Reno Bladergroen</i>
13:50	(11) Breast cancer genetic counselling referrals and counselees' knowledge, risk perception, worry and information needs in the UK and the Netherlands: How comparable are the research findings between different countries? – <i>Dr Akke Albada</i>	17:30	(24) Large cell calcified Sertoli cell tumors: differentiation between isolated and tumor syndrome cases? – <i>Dr Liesbeth Spruijt</i>
		17:40	(25) Birth incidence and prevalence of tumour prone syndromes: estimates from a UK genetic family register service <i>Prof Gareth Evans</i>
		17:50-18:00	Closing remarks: <i>Prof Anneke Lucassen and Dr Frederik Hes</i>

Programme UK/Dutch Clinical Genetics Societies Spring conference

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Thursday 11 March 2010

09:00 Registration and coffee

10:00 Opening address – *Prof Nine Knoers*

10:05-12:10 **SYMPOSIUM: Dysmorphology syndromes and Cancer/ Dysmorphology in Cancer syndromes**

Chairs: *Dr Mark Davies* and *Prof Hanne Meijers-Heijboer*

10:05 (26) Birt-Hogg-Dubé syndrome: clinical and molecular diagnosis

Dr Fred Menko

10:30 (27) RAS-MAPK pathway and cancer – *Dr Bronwyn Kerr*

10:55 (28) Dyskeratosis Congenita – *Prof Inderjeet Dokal*

SUBMITTED ABSTRACTS

11:20 (29) Evaluation and follow-up of 50 families with suspected Birt-Hogg-Dubé syndrome; a multi-center study in the Netherlands
Arjan Houweling

11:30 (30) Increased lifetime risk of cancer in patients with Noonan Syndrome carrying a PTPN11 mutation – *Marjolijn Jongmans*

11:40 (31) Genome-wide paternal uniparental disomy in a patient with Beckwith Wiedemann syndrome and multiple tumours
Dr Sixto Garcia-Minaur

11:50 (32) Neurofibromas are part of the phenotype of CDKN2A mutations that affect p14ARF transcript. A report of two Dutch families with one founder mutation – *Albertien van Eerde*

12:00 (33) Germ line WTX gene defect cause osteopathia striata with sclerosing skeletal dysplasia but does not lead to tumour development in affected individuals – *Dr Suzanna Frints*

12:10 Annual General Meeting CGS

12:40-13:30 **LUNCH and POSTER VIEWING**

13:30-16:00 **SYMPOSIUM: New technologies: the many faces of implementation**

Chairs: *Prof Sir John Burn* and *Dr Jan Oosterwijk*

13:30 (34) A dedicated SureSelect exon capture system for high throughput sequencing of the kinome in large numbers of cancer samples
Prof René Bernards (sponsored by Agilent)

14:00 (35) How next generation sequencing is changing the practice of clinical genetics – *Prof Han Brunner*

14:20 (36) UVs in cancer: making Sense out of Missense
Prof Robert Hofstra

14:40 (37) MutaDATABASE, a standardized, centralized, open access database of mutations leading to human genetic disease
Dr Patrick Willems (sponsored by Gendia)

14:50 (38) How to survive the sequence boom?
Submit your mutations and unclassified variants to databases
Prof Martijn Breuning

15:00 (39) UK point of view
New technologies; more information. What to tell patients and their relatives? – *Prof Anneke Lucassen*

15:15 (40) Dutch point of view – *Dr Rolf Sijmons*

15:30 Discussion – Lead Discussant – *Prof Anneke Lucassen*

16:00-16:30 **TEA**

16:30-18:10 **SUBMITTED ABSTRACTS**

Chairs: *Dr Frances Flinter* and *Dr Mieke van Haelst*

Robin Winter Prize presentations (UK SpRs)

16:30 (41) Genomic Loci in Agenesis of the Corpus Callosum
Dr Mary O'Driscoll

16:40 (42) Delineation of severe congenital neutropenia type4 phenotype
Dr Siddharth Banka

16:50 (43) Cantú Syndrome: Hypertrichosis, distinctive facial features, cardiomegaly and mild osteochondrodysplasia – *Dr Ingrid Scurr*

17:00 (44) DICER1 Syndrome - A New Tumour Predisposition Syndrome
Dr Ingrid Slade

17:10 (45) A clinical and molecular study of OAVS – *Dr Rachel Cole*

17:20 (46) Cryptic Chromosomal Abnormalities are a Common Cause of Syndromic Obesity: Array CGH study and review – *Dr Rob Hastings*

The Dutch SpR presentations

17:30 (47) Copy number variations of obesity genes in early-onset severe obesity – *Eelco Dulfer*

17:40 (48) CHIP-based sequence analysis of 34 cardiomyopathy genes reveals new genes involved in HCM and DCM and multiple pathogenic mutations in single patients – *Dr Ingrid Krapels*

17:50 (49) Prenatal diagnosis of MYH7 associated noncompaction cardiomyopathy – *Yvonne Hoedemaekers*

18:00 (50) The 17q21.31 microdeletion syndrome – *Dr David Koolen*

18:10-19:15 **VISIT TO THE ZOO**

19:15 **CONFERENCE DINNER**

Robin Winter prize announcement

Friday 12 March 2010

- 09:00-10:35 **SYMPOSIUM: Genetics of Brain Malformations**
Chairs: *Prof Yanick Crow* and *Dr Alice Brooks*
- 09:00 (51) Dissecting the genetic heterogeneity in Walker-Warburg syndrome and related cobblestone lissencephalies – *Dr Hans van Bokhoven*
- 09:25 (52) Mechanisms of cortical malformation – *Dr Grazia Mancini*
- 09:50 (53) Pontocerebellar hypoplasia – *Prof Peter Barth*
- SUBMITTED ABSTRACTS**
- 10:15 (54) An Atypical atypical Presentation presentation of Filamin A Mutation mutation in 2 brothers – *Renske Oegema*
- 10:25 (55) Severe encephaloclastic brain lesions in association with COL4A1-mutations – *Marije Meuwissen*
- 10:35-11:05 **COFFEE**
- 11:05-12:50 **SYMPOSIUM: Sensory systems**
Chairs: *Dr Maria Bitner-Glindzicz* and *Dr Bert de Vries*
- 11:05 (56) How not to build an eye – *Dr David Fitzpatrick*
- 11:30 (57) Towards molecular diagnostics for hereditary hearing loss with a recessive pattern of inheritance – *Dr Hannie Kremer*
- 11:55 (58) The nose – *Prof Raoul Hennekam*
- SUBMITTED ABSTRACTS**
- 12:20 (59) Multidisciplinary outpatient clinic for children with hearing loss
Dr Sarina Kant
- 12:35 (60) The expanding phenotype of CHD7 mutations
Dr Conny van Ravenswaaij-Arts
- 12:50-14:00 **LUNCH and POSTER VIEWING**
- 14:00-15:05 **SUBMITTED ABSTRACTS**
Chairs: *Dr Geoff Woods* and *Prof Nine Knoers*
- 14:00 (61) CCBE1: a new gene for recessively inherited generalised lymphatic dysplasia – *Dr Fiona Connell*
- 14:15 (62) Application of genome wide 250K SNP array analysis in prenatal diagnosis – *Dr Ineke van der Brugt*
- 14:30 (63) Unexpected findings in array analysis – *Dr Caroline Ogilvie*
- 14:50 (64) Drosophila as a model organism to establish a genotype-phenotype correlation in 2q23.1 deletion syndrome
Bregje van Bon

- 15:05-16:00 **SYMPOSIUM: Ciliopathies**
- 15:05 (65) OFD1 is mutated in X-linked Joubert syndrome and interacts with the Leber congenital amaurosis-associated protein lebercilin – *Karlien Coene*
- 15:20 (66) Lumping, splitting and treating ciliopathies – *Prof Phil Beales*
- 16:00 Conference closure – *Prof Nine Knoers*
- 16:10-17:00 **DRINKS**

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