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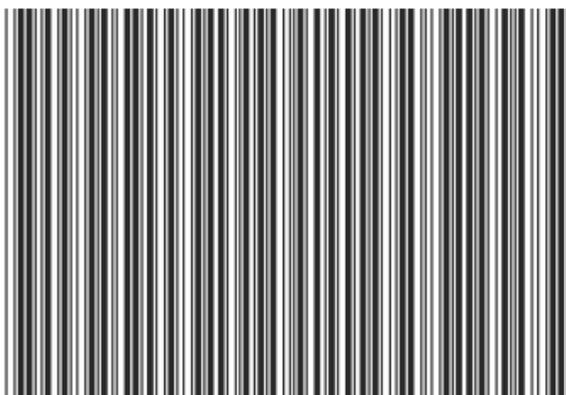
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Deze barcode geeft u toegang tot de parking van de
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Vrije Universiteit Brussel

FACULTEIT GENEESKUNDE EN FARMACIE

Doctoraat Medische Wetenschappen

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UITNODIGING

Voor de openbare verdediging van het
doctoraatsproefschrift van

An JANSEN

24 januari 2008

U wordt vriendelijk uitgenodigd op de openbare verdediging van het proefschrift van

An JANSEN

'Phenotypes and Genotypes in Symptomatic and Idiopathic Epilepsy Syndromes'

Situering van het proefschrift

We have studied the phenotypes, where possible in relation to the genotypes, in symptomatic and idiopathic epilepsy syndromes. We have expanded the phenotypes and have reviewed and elaborated the genotypes in different malformations of cortical development, including polymicrogyria and periventricular heterotopia. We have provided a more detailed description of the epilepsy phenotype in genetic disorders that are frequently associated with epilepsy, including tuberous sclerosis complex, vanishing white matter disease, Lafora disease and chorea-acanthocytosis. We have studied the genetic basis underlying idiopathic epilepsy syndromes and have provided guidelines for the selection of patients in whom genetic testing might be useful. When evaluating the contribution of genetic testing (genotype) to patient counseling and treatment (phenotype), the disorders studied in this thesis can be separated into two groups. The genetic basis of disorders in the first group, including tuberous sclerosis complex, Lafora disease, and others, has almost completely been clarified, and the determination of the genotype is starting to play a significant role in counseling, management, and treatment of patients. Disorders in the second group, including polymicrogyria, heterotopia, and most idiopathic epilepsies, are likely to result from complex inheritance, with interplay between multiple genes as well as environmental factors. Research in this group is still in the phase of gene-hunting and the current contribution of the genotype to the management of individual patients remains limited. It is hoped that the research presented in this thesis will help make a contribution to our understanding of the epilepsies and will highlight the importance of further research in this field.

Curriculum Vitae

After finishing medical school at the University of Antwerp in 1999, Dr An Jansen started her training in neurology under the supervision of Prof. Guy Ebinger at the UZ Brussel. In June 2003, she left Brussels to train with Dr Eva Andermann, who runs the Neurogenetics Unit at the Montreal Neurological Hospital and Institute, and her husband, Dr Frederick Andermann, Head of the Epilepsy Clinic. She studied the epilepsy phenotype and genotype in a variety of inherited neurological disorders including polymicrogyria, periventricular nodular heterotopia, and tuberous sclerosis complex. In January 2005, she started a two-year-training in pediatric neurology with Prof. Linda De Meirleir at the UZ Brussel. Under her supervision, and in collaboration with Willy Lissens and Sara Seneca, PhDs in the Center for Medical Genetics, she set up a project to investigate the genetic basis of malformations of cortical development. She also continues her collaboration with Prof. Peter De Jonghe and the epilepsy group at the University of Antwerp. The combination of these approaches constitutes her current opportunity to further contribute to the field of epilepsy. Her research has resulted in the publication of 19 peer-reviewed articles, 9 of which as first author, and has been presented internationally. She has won an award from the Belgische Stichting Roeping in 2003 and from the VVN in 2007. She has obtained funding from the Savoy Foundation for Epilepsy Research in 2004 and from the Scientific Fund Willy Gepts in 2007 and 2008. Dr Jansen currently works as a pediatric neurologist at the UZ Brussel.

Op **donderdag 24 januari 2008** om **17 uur** in auditorium **P. Brouwer** van de Faculteit Geneeskunde & Farmacie, Laarbeeklaan 103, 1090 Brussel