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The human genome; you gain some, you lose some

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CURRICULUM VITAE

Naam: Marjolein Kriek
Geboortedatum: 22-11-1973
Geboorte plaats: Leiden (Academisch Ziekenhuis Leiden)

School

Eindexamen atheneum aan het Visser 't Hooft lyceum te Leiden (1992).

Studies

- 18 August 2000
Behalen van de artsenbul aan de Universiteit Leiden.
- 17 september 2002
Doctoraal examen van de studie Biomedische Wetenschappen aan de Universiteit Leiden.

Wetenschappelijk onderzoek

1995 Zes maanden stage bij vakgroep Moleculaire Carcinogenese aan Universiteit Leiden o.l.v. Prof. Dr van der Eb en Dr Zantema.

Titel onderzoek:

“Association of proteins influenced by the Adenovirus E1A oncoprotein”.

1998 Drie maanden stage bij vakgroep Klinische Epidemiologie in het L.U.M.C. o.l.v. Prof. Dr Roosendaal en Drs Sramek.

Titel onderzoek:

“Mortality in carriers of Hemophilia”.

Dit onderzoek leidde tot een tweede auteurschap in de Lancet.

2001 Eindvakstage Biomedische Wetenschappen (9 maanden) bij de vakgroep Humane en Klinische Genetica o.l.v. Prof. Breuning

Titel onderzoek:

“Screening for mutations in mentally retarded patients using MAPH”.

Dit onderzoek vormde de basis van het huidige proefschrift.

2002 Begonnen aan promotie onderzoek getiteld; “The human genome; you gain some, you lose some”, onder leiding van Prof. M.H.Breuning, Prof. G-J B. Van Ommen en dr. J.T. den Dunnen: Aanvankelijk als AGNIO, vanaf 1 januari 2003 is dit omgezet in een AGIKO traject op basis van ZONMW-subsidie (AGIKO-fellowship 940-37-032).

Klinische ervaring

2000 Half jaar als AGNIO gewerkt op de afdeling Klinische Genetica (LUMC)

1 april 2005 tot heden

In opleiding tot klinisch geneticus op de afdeling Klinische Genetica (LUMC)

LIST OF PUBLICATIONS**2002**

White S, Kalf M, Liu Q, Villerius M, Engelsma D, Kriek M, Vollebregt E, Bakker B, van Ommen GJ, Breuning MH *et al.* Comprehensive detection of genomic duplications and deletions in the DMD gene, by use of multiplex amplifiable probe hybridization. *Am J Hum Genet.* 2002 Aug;71(2):365-74.

2003

Sramek A, Kriek M, Rosendaal FR. Decreased mortality of ischaemic heart disease among carriers of haemophilia. *Lancet.* 2003 Aug 2;362(9381):351-4

2004

Kriek M, White SJ, Bouma MC, Dauwerse HG, Hansson KB, Nijhuis JV, Bakker B, van Ommen GJ, den Dunnen JT, Breuning MH. Genomic imbalances in mental retardation. *J Med Genet.* 2004 Apr;41(4):249-55

White SJ, Vink GR, Kriek M, Wuyts W, Schouten J, Bakker B, Breuning MH, den Dunnen JT. Two-color multiplex ligation-dependent probe amplification: detecting genomic rearrangements in hereditary multiple exostoses. *Hum Mutat.* 2004 Jul;24(1):86-92.

2006

Rosenberg C, Knijnenburg J, Bakker E, Vianna-Morgante AM, Sloos W, Otto PA, Kriek M, Hansson K, Krepischi-Santos AC, Fiegler H, Carter NP, Bijlsma EK, van Haeringen A, Szuhai K, Tanke HJ. Array-CGH detection of micro rearrangements in mentally retarded individuals: clinical significance of imbalances present both in affected children and normal parents. *J Med Genet.* 2006 Feb;43(2):180-6.

Kriek M, White SJ, Szuhai K, Knijnenburg J, van Ommen GJ, den Dunnen JT, Breuning MH. Copy number variation in regions flanked (or unflanked) by duplicons among patients with developmental delay and/or congenital malformations; detection of reciprocal and partial Williams-Beuren duplications.

Eur J Hum Genet. 2006 Feb;14(2):180-9

van der Knaap MS, Kriek M, Overweg-Plandsoen WC, Hansson KB, Madan K, Starreveld JS, Schotman-Schram P, Barkhof F, Lesnik Oberstein SA. Cerebral white matter abnormalities in 6p25 deletion syndrome.

AJNR Am J Neuroradiol. 2006 Mar;27(3):586-8

Kriek M, Szuhai K, Kant SG, White SJ, Dauwerse H, Fiegler H, Carter NP, Knijnenburg J, den Dunnen JT, Tanke HJ, Breuning MH, Rosenberg C. A complex rearrangement on chromosome 22 affecting both homologues; haplo-insufficiency of the Cat eye syndrome region may have no clinical relevance.

Hum Genet. 2006 Aug;120(1):77-84.

Lesnik Oberstein SA, Kriek M, White SJ, Kalf ME, Szuhai K, den Dunnen JT, Breuning MH, and Hennekam RC. Peters Plus Syndrome Is Caused by Mutations in *B3GALTL*, a Putative Glycosyltransferase.

Am J Hum Genet. 2006 Aug; 79(3):562-6.

Rosenberg C, Krepischi-Santos ACV, Knijnenburg J, Kok F, Otto PA, Tanke HJ, Kriek M, Zangrande Vieira LC, Nascimento RMP, Vianna-Morgante AM. X-chromosome segmental imbalances as a cause of recessive mental retardation syndromes.

J Med Genet. 2006 Feb;43(2):180-6.

2007

Kant SG, Kriek M, Walenkamp MJE, Hansson KBM, van Rhijn A, Clayton-Smith J, Wit JM, Breuning MH. Tall stature and duplication of the insulin-like growth factor I receptor gene.

Eur J Med Genet. 2007 Jan-Feb;50(1):1-10.

Kriek M, Konijnenburg J, White SJ, Rosenberg C, den Dunnen JT, van Ommen GJ, Tanke HJ, Breuning MB, Szuhai K. Diagnosis of genetic abnormalities in developmentally delayed patients: A new strategy combining MLPA and Array-CGH.

Am J Med Genet A. 2007 Mar 15;143(6):610-4.

Harteveld CL, Kriek M, Bijlsma EK, Erjavec Z, Balak D, Phylipsen M, Voskamp A, di Capua E, White SJ and Giordano PC. Telomeric deletions of 16p causing alpha-thalassemia and mental retardation characterized by multiplex ligation-dependent probe amplification.

Human Genet. 2007 Jun 28; [Epub ahead of print]

Kriek M, Ruivenkamp CAL, Ariyurek Y, Kalf ME, Konijnenburg J, van Haeringen A, Genuardi M, Rosenberg C, Sanders SR., White SJ, Szuhai K, Breuning MH, den Dunnen JT. Comparison of four genome-wide platforms using overlapping interstitial 2p alterations.

Submitted

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