Advanced genome-wide screening in human genomic disorders

Knijnenburg, J.

Curriculum vitae

Jeroen Knijnenburg (27 mei 1977, Borculo) behaalde het VWO diploma in 1995 aan het Staring College te Lochem en het HBO diploma (HLO, Hogere Laboratorium Opleiding, richting Biochemie) in 1999 aan de Rijkshogeschool IJselland te Deventer. In het laatste jaar van de opleiding heeft hij zijn stageperiode vervuld op de afdeling Moleculaire Celbiologie van Pharming Technologies B.V. te Leiden, onder de leiding van drs. G. Platenburg en dr. J.J. Heus. Vanaf juni 1999 is hij ruim twee jaar werkzaam geweest als analist op dezelfde afdeling van Pharming Technologies B.V. te Leiden.
Vanaf oktober 2001 was hij werkzaam bij het Leids Universitair Medisch Centrum op de afdeling Moleculaire Celbiologie van prof. dr. H.J. Tanke, in eerste instantie onder de leiding van dr. C. Rosenberg, later onder de leiding van dr. K. Szuhai. In maart 2005 is hij begonnen als promovendus op dezelfde afdeling onder de leiding van dr. K. Szuhai, met als onderzoeksdoel structurele genetische afwijkingen op te sporen en te karakteriseren bij patiënten met mentale retardatie met het eerder opgezette array-CGH platform.
Sinds januari 2009 is Jeroen Knijnenburg werkzaam bij de afdeling Klinische Genetica (prof. dr. F.G. Grosveld) van het Erasmus Medisch Centrum te Rotterdam, alwaar hij een opleiding volgt tot Klinisch Cytogeneticus.

List of publications

1. Raap AK, van der Burg MJ, Knijnenburg J, Meershoek E, Rosenberg C, Gray JW, Wiegant J, Hodgson JG, Tanke HJ. Array comparative genomic hybridization with cyanin cis-platinum-labeled DNAs. Biotechniques 2004 Jul;37(1):130-4.
2. Szuhai K, Knijnenburg J, IJszenga M, Tanke HJ, Baatenburg de Jong RJ, Bas Douwes DP, Rosenberg C, Hogendoorn PC. Multicolor fluorescence in situ hybridization analysis of a synovial sarcoma of the larynx with a t(X;18)(p11.2;q11.2) and trisomies 2 and 8. Cancer Genet Cytogenet 2004 Aug;153(1):48-52.
3. Knijnenburg J, Szuhai K, Giltay J, Molenaar L, Sloos W, Poot M, Tanke HJ, Rosenberg C. Insights from genomic microarrays into structural chromosome rearrangements. Am J Med Genet A 2005 Jan 1;132(1):36-40.
4. Rosenberg C, Knijnenburg J, Chauffaille ML, Brunoni D, Catelani AL, Sloos W, Szuhai K, Tanke HJ. Array CGH detection of a cryptic deletion in a complex chromosome rearrangement. Hum Genet 2005 Apr;116(5):390-4.
5. Jehee FS, Rosenberg C, Krepischi-Santos AC, Kok F, Knijnenburg J, Froyen G, Vianna-Morgante AM, Opitz JM, Passos-Bueno MR. An Xq22.3 duplication detected by comparative genomic hybridization microarray (Array-CGH) defines a new locus (FGS5) for FG syndrome. Am J Med Genet A 2005 Dec 15;139(3):221-6.
6. Knijnenburg J, van der Burg M, Nilsson P, Ploos van Amstel HK, Tanke H, Szuhai K. Rapid detection of genomic imbalances using micro-arrays consisting of pooled BACs covering all human chromosome arms. Nucleic Acids Res 2005;33(18):e159.
7. Krepischi-Santos AC, Vianna-Morgante AM, Jehee FS, Passos-Bueno MR, Knijnenburg J, Szuhai K, Sloos W, Mazzeu JF, Kok F, Cheroki C, Otto PA, Mingroni-Netto RC, Varela M, Koiffmann C, Kim CA, Bertola DR, Pearson PL, Rosenberg C. Whole-genome array-CGH screening in undiagnosed syndromic patients: old syndromes revisited and new alterations. Cytogenet Genome Res 2006;115(3-4):254-61.
8. Varela MC, Krepischi-Santos AC, Paz JA, Knijnenburg J, Szuhai K, Rosenberg C, Koiffmann CP. A 17q21.31 microdeletion encompassing the MAPT gene in a mentally impaired patient. Cytogenet Genome Res 2006;114(1):89-92.
9. Dijkman R, Tensen CP, Jordanova ES, Knijnenburg J, Hoefnagel JJ, Mulder AA, Rosenberg C, Raap AK, Willemze R, Szuhai K, Vermeer MH. Array-based comparative genomic hybridization analysis reveals recurrent chromosomal alterations and prognostic parameters in primary cutaneous large B-cell lymphoma. J Clin Oncol 2006 Jan 10;24(2):296-305.
10. 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 HA, 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.
11. 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.
12. 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.
13. Kloth JN, Oosting J, van Wezel T, Szuhai K, Knijnenburg J, Gorter A, Kenter GG, Fleuren GJ, Jordanova ES. Combined array-comparative genomic hybridization and single-nucleotide polymorphism-loss of heterozygosity analysis reveals complex genetic alterations in cervical cancer. BMC Genomics 2007;8:53.

14. Hoffer MJ, Hilhorst-Hofstee Y, Knijnenburg J, Hansson KB, Engelberts AC, Laan LA, Bakker E, Rosenberg C. A 6Mb deletion in band 2q22 due to a complex chromosome rearrangement associated with severe psychomotor retardation, microcephaly and distinctive dysmorphic facial features. Eur J Med Genet 2007 Mar;50(2):149-54.
15. Kriek M, Knijnenburg J, White SJ, Rosenberg C, den Dunnen JT, van Ommen GJ, Tanke HJ, Breuning MH, 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.
16. Hameetman L, Szuhai K, Yavas A, Knijnenburg J, van DM, van DH, Taminiau AH, Cleton-Jansen AM, Bovee JV, Hogendoorn PC. The role of EXT1 in nonhereditary osteochondroma: identification of homozygous deletions. J Natl Cancer Inst 2007 Mar 7;99(5):396-406.
17. Knijnenburg J, van Haeringen A, Hansson KB, Lankester A, Smit MJ, Belfroid RD, Bakker E, Rosenberg C, Tanke HJ, Szuhai K. Ring chromosome formation as a novel escape mechanism in patients with inverted duplication and terminal deletion. Eur J Hum Genet 2007 May;15(5):548-55.
18. Hansson K, Szuhai K, Knijnenburg J, van Haeringen A, de Pater J. Interstitial deletion of 6q without phenotypic effect. Am J Med Genet A 2007 Jun 15;143(12):1354-7.
19. Szuhai K, IJszenga M, Knijnenburg J, Dijkstra S, de SA, Tanke HJ, Hogendoorn PC. Does parosteal liposarcoma differ from other atypical lipomatous tumors/well-differentiated liposarcomas? A molecular cytogenetic study using combined multicolor COBRA-FISH karyotyping and array-based comparative genomic hybridization. Cancer Genet Cytogenet 2007 Jul 15;176(2):115-20.
20. Mazzeu JF, Krepischi-Santos AC, Rosenberg C, Szuhai K, Knijnenburg J, Weiss JM, Kerkis I, Mustacchi Z, Colin G, Mombach R, Pavanello RC, Otto PA, Vianna-Morgante AM. Chromosome abnormalities in two patients with features of autosomal dominant Robinow syndrome. Am J Med Genet A 2007 Aug 1;143(15):1790-5.
21. Knijnenburg J, van der Burg M, Tanke HJ, Szuhai K. Optimized amplification and fluorescent labeling of small cell samples for genomic array-CGH. Cytometry A 2007 Aug;71(8):585-91.
22. Vermeer MH, van Doorn R, Dijkman R, Mao X, Whittaker S, van Voorst Vader PC, Gerritsen MJ, Geerts ML, Gellrich S, Soderberg O, Leuchowius KJ, Landegren U, Out-Luiting JJ, Knijnenburg J, IJszenga M, Szuhai K, Willemze R, Tensen CP. Novel and highly recurrent chromosomal alterations in Sezary syndrome. Cancer Res 2008 Apr 15;68(8):2689-98.
23. Van Doorn R, van Kester MS, Dijkman R, Vermeer MH, Mulder AA, Szuhai K, Knijnenburg J, Boer JM, Willemze R, Tensen CP. Oncogenomic analysis of mycosis fungoides reveals major differences with Sezary syndrome. Blood. 2008 Oct 1 (*Epub ahead of print*).
24. Goobie S, Knijnenburg J, FitzPatrick D, Sharkey FH, Christopher Lionel A, Marshall CR, Azam T, Shago M, Chong K, Mendoza-Londono R, den Hollander NS, Ruivenkamp CAL, Maher E, Tanke HJ, Szuhai K, Wintle RF, Scherer SW. Molecular and clinical characterization of de novo and familial cases with microduplication 3q29: guidelines for copy number variation case reporting. Cytogenet Genome Res 2008 Dec;123: (DOI:10.1159/000184693).
25. Knijnenburg J, Lesnik Oberstein SAJ, Frei K, Lucas T, Gijsbers ACJ, Ruivenkamp CAL, Tanke HJ, Szuhai K. A homozygous deletion of a normal variation locus causes syndromic hearing loss in a patient from non-consanguineous parents. J Med Genet 2008, *submitted*.
26. Aten E, den Hollander NS, Ruivenkamp CAL, Knijnenburg J, Van Bokhoven H, Den Dunnen JT, Breuning MH. Split Hand-Foot Malformation, Fallot's tetralogy, Angelman like phenotype and a 1Mb 19p deletion – evidence for further heterogeneity? Am J Med Genet 2009, *in press*.
27. Unger K, Wienberg J, Riches A, Hieber L, Walch A, Brown A, O'Brien PCM, Briscoe C, Gray L, Rodriguez E, Jackl G, Knijnenburg J, Tallini G, Ferguson-Smith M, Zitzelsberger H. Novel gene rearrangements in breast carcinogenesis identified by array-painting analysis of chromosomal aberrations. Genes Chromosomes Cancer 2008, *submitted*.
28. Lugtenberg D, Zangrande-Vieira L, Kirchhoff M, Kjaergaard S, Vianna-Morgante AM, Kleefstra T, Ruiter M, Jehee FS, Ulmann R, Veltman J, Vrijenhoek T, Pfundt R, Hoeijmakers J, Hehir-Kwa J, Oudakker AR, Chelly J, Froyen G, Ropers HH, Moraine C, Gecz J, Knijnenburg J, Kant SG, Hamel BCJ, Van Bokhoven H, Rosenberg C, De Brouwer APM. A recurrent deletion of *ZNF630* in Xp11.23: a risk factor for mental retardation? BMC Genomics 2008, *submitted*.