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Rolf H. Sijmons, MD, PhD



Professor of Medical Translational Genetics, Head of Development and Innovation section



Prof. Rolf Sijmons

MD: Medicine, University of Groningen, 1986

Specialist qualification in clinical genetics, 1993, Groningen

PhD: Studies on hereditary cancer, University of Groningen, 1999

Inaugural lecture: "Genomic Medicine: een toekomstige zorg?" [Full text](#) (in Dutch), [cover](#), 3 December 2013

News:

2015: 5GPM - Five genes per minute - Rapid whole genome sequencing for critically ill newborns [more](#)

2014: NTR Radio interview on Genomic Medicine ([listen](#), in Dutch, first item, time slot 3.00-08.50 mins)

2013: [Better understanding of DNA findings](#) in Familial Bowel Cancer

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See [Google Scholar](#) or [PubMed](#) for his publications

Keywords

Clinical applications of next-generation technology, Familial and hereditary cancer, Lynch syndrome, MMR genes, applied informatics in medical genetics

Short cv

Rolf Sijmons is a consultant clinical geneticist and professor of medical translational genetics at the Department of Genetics, UMCG. He heads the department's [Development & Innovation section](#), which aims to translate new knowledge and technology into clinical practice. This section also includes the oncogenetics research team. See also [Systems Genetics](#).

His main clinical and research interests are familial and hereditary cancer, the clinical application of next-generation technology, and applied informatics in medical genetics. He is the organizer of the nationally accredited UMCG expert centre on familial colorectal cancer and a member of the EU accredited European Reference Network (ERN) on hereditary cancer (GENTURIS).

He is one of the curators of the international InSiGHT database on mismatch repair (MMR) gene mutations (<http://www.insight-group.org>) and a member of the InSiGHT MMR UV interpretation committee. He is also the editor of the [Familial Cancer Database](#), a communicating editor for [Human Mutation](#), and one of the three editors-in-chief of [Hereditary Cancer in Clinical Practice](#).

Some recent papers

- Van der Velde KJ, de Boer EN, van Diemen CC, Sikkema-Raddatz B, Abbott KM, Knopperts A, Franke L, Sijmons RH, de Koning TJ, Wijmenga C, Sinke RJ, Swertz MA. [GAVIN: Gene-Aware Variant Interpretation for medical sequencing](#). Genome Biol. 2017;18(1):6.
- Sollie A, Sijmons RH, Helsper C, Numans ME. [Reusability of coded data in the primary care electronic medical record: A dynamic cohort study concerning cancer diagnoses](#). Int J Med Inform. 2017;99:45-52.
- Sollie A, Roskam J, Sijmons RH, Numans ME, Helsper CW. [Do GPs know their patients with cancer? Assessing the quality of cancer registration in Dutch primary care: a cross-sectional validation study](#). BMJ Open. 2016;6(9): e012669 6.
- Sijmons RH, Hofstra RM. [Clinical aspects of hereditary DNA Mismatch repair gene mutations](#). DNA Repair (Amst). 2016;38:155-62
- Johansson LF, van Dijk F, de Boer EN, van Dijk-Bos KK, Jongbloed JD, van der Hout AH, Westers H, Sinke RJ, Swertz MA, Sijmons RH, Sikkema-Raddatz B. [CoNVaDING: Single Exon Variation Detection in Targeted NGS Data](#). Hum Mutat. 2016;37(5):457-64. doi: 10.1002/humu.22969.
- Thompson BA, ... Sijmons R, ... on behalf of InSiGHT. [Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database](#). Nat Genet. 2013; doi: 10.1038/ng.2854. [abstract](#)
- Sikkema-Raddatz B, Johansson LF, ..., Sijmons RH, Jongbloed JD, Sinke RJ. [Targeted next-generation sequencing can replace cancer sequencing in](#)

[targeted next-generation sequencing can replace Sanger sequencing in clinical diagnostics](#). *Hum Mutat.* 2013;34(7):1035-42.

- Plazzer JP, Sijmons RH, et al. [The InSiGHT database: utilizing 100 years of insights into Lynch syndrome](#). *Fam Cancer.* 2013;12(2):175-80.
- Herkert JC, Niessen RC, Olderoode-Berends MJW, ..., Rolf H. Sijmons. [Paediatric intestinal cancer and polyposis due to bi-allelic PMS2 mutations: Case series, review and follow-up guidelines](#), *European J Cancer*, 2011; 47(7):965-82

Sijmons RH, van Langen IM, Sijmons JG. [A clinical perspective on ethical issues in genetic testing](#). *A ccountability in Research* 2011; 18:148-162. doi: 10.1080/08989621.2011.575033.

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