

nieuws rond erfelijk colorectaal carcinoom

Rolf Sijmons

klinisch geneticus, hoogleraar medische translationele genetica

Afdeling Genetica, UMCG

r.h.sijmons@umcg.nl

- InSiGHT



- EHTG



- GENTURIS



European
Reference
Networks

International Society for Gastrointestinal Hereditary Tumours (InSiGHT)

www.insight-group.org

News

Contact us

[Make a donation >](#)

[Become a member >](#)

Already a member? [Login](#)

[Home](#)

[About us](#)

[Meetings](#)

[Syndromes](#)

[Variants](#)

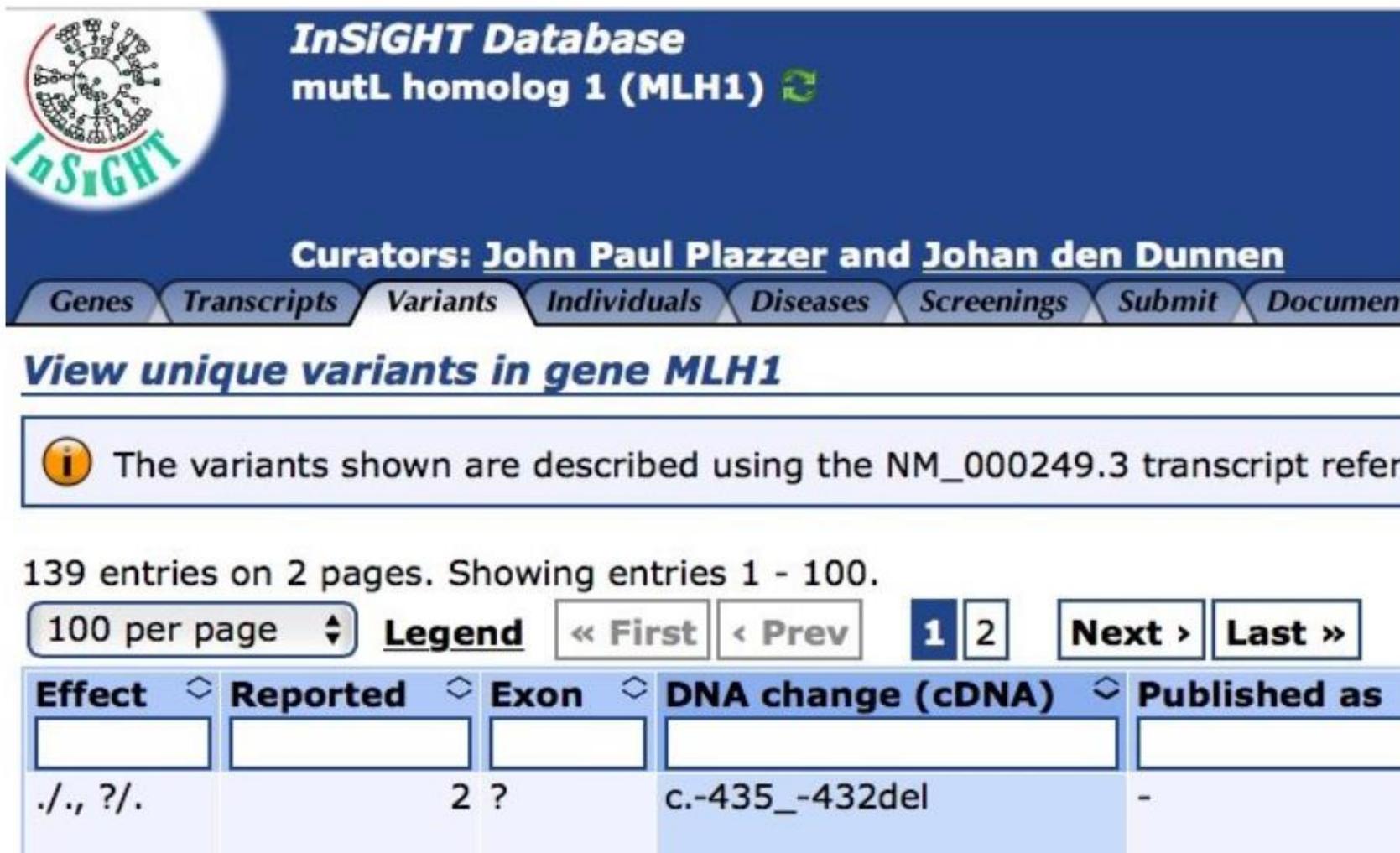
[Research](#)

The International Society for Gastrointestinal Hereditary Tumours (InSiGHT) is an international multidisciplinary, scientific organisation. Our aim is to improve the quality of care of patients and families with any hereditary condition resulting in gastrointestinal tumours



InSiGHT variant databases

InSiGHT houses and curates the most comprehensive database of DNA variants re-sequenced in the genes that contribute to gastrointestinal cancer.



The screenshot shows the InSiGHT Database interface for the **mutL homolog 1 (MLH1)** gene. The top navigation bar includes links for **Genes**, **Transcripts**, **Variants** (which is the active tab), **Individuals**, **Diseases**, **Screenings**, **Submit**, and **Documents**. A banner at the top states: **Curators: John Paul Plazzer and Johan den Dunnen**. Below this, a blue header bar features the InSiGHT logo and the text: **View unique variants in gene MLH1**. A note in a yellow box indicates: **i The variants shown are described using the NM_000249.3 transcript reference**. The main content area displays a table with 139 entries, showing columns for Effect, Reported, Exon, DNA change (cDNA), and Published as. The first entry in the table is: **./, ?/.**, **2 ?**, **c.-435_-432del**, and **-**.

Een van de
InSiGHT taken:

“lastige”
genvarianten
(VUS) worden in
groepsverband
klinisch
geclassificeerd

Wereldwijd nr 1
voor de Lynch
syndroom genen



AUCKLAND, NEW ZEALAND

**INTERNATIONAL SOCIETY FOR
GASTROINTESTINAL HEREDITARY
TUMOURS (InSiGHT)**

20 – 23 March 2019





European Hereditary Tumour Group

www.ehtg.org

2019
Barcelona, Sept
voor of na ESMO

PLSD
The Prospective Lynch Syndrome Database

ABOUT US

3RD MEETING 2018

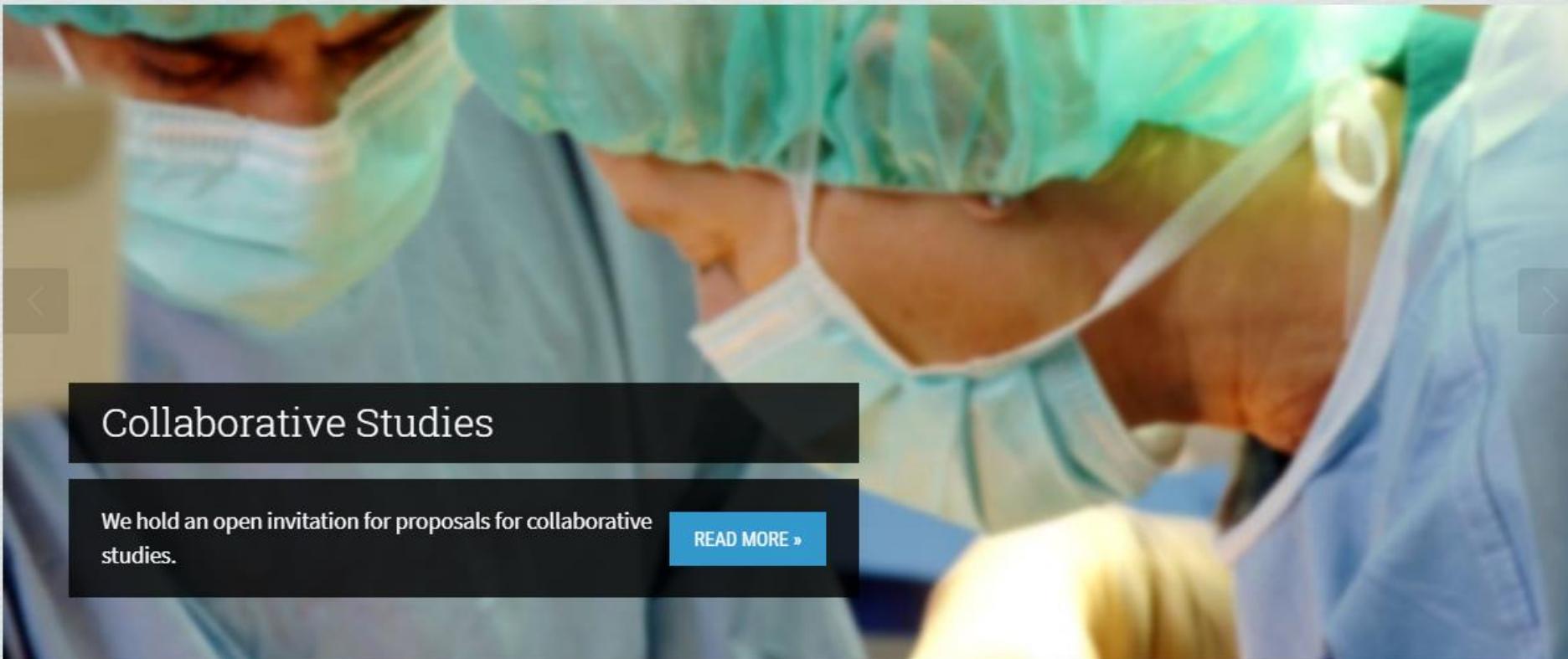
PAST MEETINGS

COLLABORATIVE STUDIES

GUIDELINES

PUBLICATIONS

MEMBERSHIP



Welcome

Become a Member of EHTG

Email Updates



Chair
Gabriela Moeslein
Surgeon

- Surgery and endoscopy
- Gynaecology
- Immunotherapy
- Chemoprevention
- MMR-Group
- Communication
- PLSD (Lynch syndrome cancer risk database)



GENTURIS is het European Reference Network (ERN) voor erfelijke kanker

focus = patiëntenzorg (consultatieve functie)



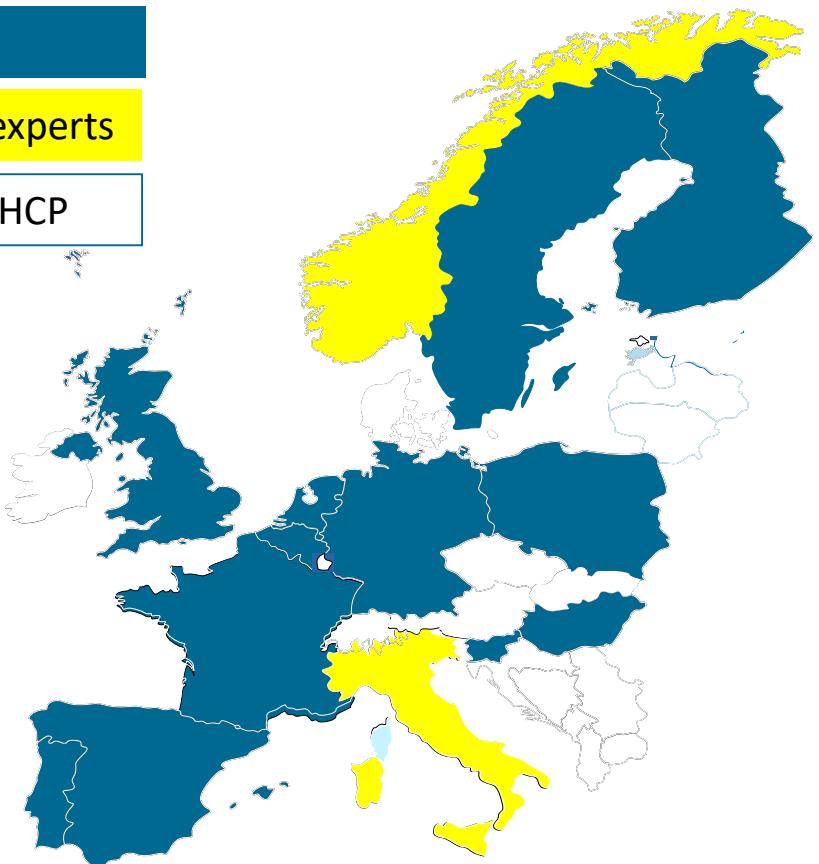
 European
Reference
Networks



ERN: Genetic Tumour Risk Syndromes

Expertise centers (full members)

- Member state with full member
- Member state with collaborating experts
- Member state without GENTURIS HCP



Member state	n =
Belgium	3
Finland	1
France	3
Germany	3
Hungary	1
Netherlands	3
Poland	1
Portugal	1
Slovenia	1
Spain	2
Sweden	1
United Kingdom	3
total	23

Thematic groups



1. Neurofibromatosis

- NF1, NF2, Schwannomatosis

2. Lynch syndrome and polyposis

- Lynch Syndrome,
- (attenuated) familial adenomatous polyposis
- MYH-Associated Polyposis, DNA polymerase proofreading associated-polyposis
- NTLH1 associated polyposis,
- Juvenile Polyposis Syndrome, Peutz-Jeghers syndrome
- Hereditary mixed polyposis, Serrated polyposis syndrome

3. Hereditary breast and ovarian cancer

4. Other rare – predominantly malignant- genturis (to be expanded)

- PTEN hamartoma tumour syndromes
- Li-Fraumeni Syndrome
- Birt-Hogg-Dubé Syndrome
- FAMMM
- Small Cell Carcinoma of the Ovary, Hypercalcaemic Type
- Hereditary diffuse gastric cancer (CDH1)



EUROPEAN SOCIETY OF HUMAN GENETICS



Spring Course in Hereditary Cancer Genetics

24th – 27th of April 2018

University Residential Centre of
Bertinoro, Italy

85 participants from 20 European countries and 12 countries outside Europe

Director of the course: N Hoogerbrugge (NL)

Organizing Committee: C Oliveira (PT), H Høberg-Vetti (NO), E Holinski-Feder (DE), together with
J Bazzoli (IT) and G Romeo (IT)

21 Interactive plenary lectures

5 Concurrent workshops in small groups

2 Poster discussion sessions and **2** Quiz sessions

Upcoming

ESMO Preceptorship on Hereditary Cancer Genetics

Lugano, 26-27 april 2019

GENTURIS symposium, t.b.a.