

2026

Effect of a polygenic risk score in patients with late-onset, early-onset, familial, or hereditary colorectal cancer.
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Mutational Landscape of Colorectal Tumors From Individuals With Unexplained Adenomatous or Serrated Colorectal Polyposis.

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Identifying Risk Factors for Metachronous Colorectal Cancer in Lynch Syndrome.

Hüneburg R, Weber K, Aretz S, Steinke-Lange V, Holinski-Feder E, Kloor M, Redler S, Vangala DB, Schröck E, Block A, Ripperger T, van Beekum K, Link A, Schroeder C, Engel C, Nattermann J; German Consortium for Familial Intestinal Cancer.

<https://pubmed.ncbi.nlm.nih.gov/40749813/>

2025

Improving genetic diagnosis of hereditary tumor syndromes: From expanded gene panels to functional genomics.

Sauer M, Lucas MC, Prokosch V, Keßler T, Risch T, Laner A, Henkel J, Benet-Pagès A, Hallermayr A, Steinke-Lange V, Holinski-Feder E, Klink B.

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Identifying risk factors for metachronous colorectal cancer in Lynch syndrome.

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Cancer prognosis and treatment results in patients with PTEN Hamartoma Tumour Syndrome (PHTS)-a European cohort study.

Hendricks LAJ, Verbeek KCJ, Schuurs-Hoeijmakers JHM, de Putter R, Brems H, Van Daele SH, Anastasiadou VC, Foretová L, Benusiglio PR, Gerasimenko A, Colas C, Villy MC, Houdayer C, Branchaud M, Hüneburg R, Aretz S, Jahn A, Steinke-Lange V, Innella G, Turchetti D, Barili V, Genuardi M, Panfili A, Baldassarri M, Irmejs A, de Jong MM, Links TP, Leter EM, Bosch DGM, Donze SH, van der Post RS, Mensenkamp AR, Westdorp H, Høberg-Vetti H, Tveit Haavind M, Jørgensen K, Mæhle L, Briskemyr S, Garcia JD, Blatnik A, Balmaña J, Torres M, Brunet J, Lleuger-Pujol R, Tham E, Tischkowitz M, Evans DG, Hyder Z, Hoogerbrugge N, Vos JR.

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A series of reviews in familial cancer: genetic cancer risk in context variants of uncertain significance in MMR genes: which procedures should be followed?

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High amount of fertility reducing tumors and procedures, but no evidence for premature ovarian failure in female Lynch syndrome patients.

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Lifestyle Factors and Breast Cancer in Females with PTEN Hamartoma Tumor Syndrome (PHTS).

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An interconnected data infrastructure to support large-scale rare disease research.

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The utility of liquid biopsy in clinical genetic diagnosis of cancer and monogenic mosaic disorders.

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Combined loss of CDH1 and downstream regulatory sequences drive early-onset diffuse gastric cancer and increase penetrance of hereditary diffuse gastric cancer.

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Wnt genes in colonic polyposis predisposition.

Quintana I, Terradas M, Mur P, Te Paske IBAW, Peters S, Spier I, Steinke-Lange V, Maestro C, Torrents D, Puiggròs M, Royo R, Tonda R, Parra G, Piscia D, Beltrán S, Navarro M, Piñol V, Brunet J, Gonzalez-Abuin N, Aiza G, Sommer A, van Herwaarden Y, Astuti G, Holinski-Feder E, Hoogerbrugge N, de Voer RM, Aretz S, Capellá G, Valle L.
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Diagnostic yield and clinical relevance of expanded germline genetic testing for nearly 7000 suspected HBOC patients.

Jan Henkel, Andreas Laner, Melanie Locher, Tobias Wohlfrom, Birgit Neitzel, Kerstin Becker, Teresa Neuhann, Angela Abicht, Verena Steinke-Lange, Elke Holinski-Feder.

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Mortality by age, gene and gender in carriers of pathogenic mismatch repair gene variants receiving surveillance for early cancer diagnosis and treatment: a report from the prospective Lynch syndrome database.

Mev Dominguez-Valentin, Saskia Haupt, Toni T Seppälä, Julian R Sampson, Lone Sunde, Inge Bernstein, Mark A Jenkins, Christoph Engel, Stefan Aretz, Maartje Nielsen, Gabriel Capella, Francesc Balaguer, Dafydd Gareth Evans, John Burn, Elke Holinski-Feder, Lucio Bertario, Bernardo Bonanni, Annika Lindblom, Zohar Levi, Finlay Macrae, Ingrid Winship, John-Paul Plazzer, Rolf Sijmons, Luigi Laghi, Adriana Della Valle, Karl Heinemann, Tadeusz Dębniak, Robert Fruscio, Francisco Lopez-Koestner, Karin Alvarez-Valenzuel, Lior H Katz, Ido Laish, Elez Vainer, Carlos Vaccar, Dirce Maria Carrar, Kevin Monahan, Elizabeth Half, Aine Stakelum, Des Winter, Rory Kennelly, Nathan Gluck, Harsh Sheth, Naim Abu-Freha, Marc Greenblatt, Benedito Mauro Rossi, Mabel Bohorquez, Giulia Martina Cavestro, Leonardo S Lino-Silva, Karoline Horisberger, Maria Grazia Tibiletti, Ivana do Nascimento, Huw Thomas, Norma Teresa Rossi, Leandro Apolinário da Silva, Attila Zaránd, Juan Ruiz-Bañobre, Vincent Heuveline, Jukka-Pekka Mecklin, Kirsi Pylvänäinen, Laura Renkonen-Sinisaal, Anna Lepistö, Päivi Peltomäki, Christina Therkildsen, Mia Gebauer Madsen, Stefan Kobelgaard Burgdorf, John L Hopper, Aung Ko Win, Robert W Haile, Noralane Lindor, Steven Gallinger, Loïc Le Marchand, Polly A Newcomb, Jane Figueiredo, Daniel D Buchanan, Stephen N Thibodeau, Magnus von Knebel Doeberitz, Markus Loeffler, Nils Rahner, Evelin Schröck, Verena Steinke-Lange, Wolff Schmiegell, Deepak Vangala, Claudia Perne, Robert Hüneburg, Silke Redler, Reinhard Büttner, Jürgen Weitz, Marta Pineda, Nuria Duenas, Joan Brunet Vidal, Leticia Moreira, Ariadna Sánchez, Eivind Hovig, Sigve Nakken, Kate Green, Fiona Laloo, James Hill, Emma Crosbie, Miriam Mints, Yael Goldberg, Douglas Tjandra, Sanne W Ten Broeke, Revital Kariv, Guy Rosner, Suresh H Advani, Lidiya Thomas, Pankaj Shah, Mithun Shah, Florencia Neffa, Patricia Esperon, Walter Pavicic, Giovana Tardin Torrezan, Thiago Bassaneze, Claudia Alejandra Martin, Gabriela Moslein, Pål Moller.

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Highly sensitive liquid biopsy Duplex sequencing complements tissue biopsy to enhance detection of clinically relevant genetic variants.

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Transcript capture and ultradeep long-read RNA sequencing (CAPLRseq) to diagnose HNPCC/Lynch syndrome.

Schwenk V, Leal Silva RM, Scharf F, Knaust K, Wendlandt M, Häusser T, Pickl JMA, Steinke-Lange V, Laner A, Morak M, Holinski-Feder E, Wolf DA. J Med Genet. 2023 Jan 2:jmg-2022-108931. doi: 10.1136/jmg-2022-108931. Online ahead of print. PMID: 36593122

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2022

Genotype-first approach to identify associations between CDH1 germline variants and cancer phenotypes: a multicentre study by the European Reference Network on Genetic Tumour Risk Syndromes

José Garcia-Pelaez, Rita Barbosa-Matos, Silvana Lobo, Alexandre Dias, Luzia Garrido, Sérgio Castedo, Sónia Sousa, Hugo Pinheiro, Liliana Sousa, Rita Monteiro, Joaquin J Maqueda, Susana Fernandes, Fátima Carneiro, Nádia Pinto, Carolina Lemos, Carla Pinto, Manuel R Teixeira, Stefan Aretz, Svetlana Bajalica-Lagercrantz, Judith Balmaña, Ana Blatnik, Patrick R Benusiglio, Maud Blanluet, Vicent Bours, Hilde Brems, Joan Brunet, Daniele Calistri, Gabriel Capellá, Sergio Carrera, Chrystelle Colas, Karin Dahan, Robin de Putter, Camille Desseignés, Elena Domínguez-Garrido, Conceição Egas, D Gareth Evans, Damien Feret, Eleanor Fewings, Rebecca C Fitzgerald, Florence Coulet, María Garcia-Barcina, Maurizio Genuardi, Lisa Golmard, Karl Hackmann, Helen Hanson, Elke Holinski-Feder, Robert Hüneburg, Mateja Krajc, Kristina Lagerstedt-Robinson, Conxi Lázaro 25, Marjolijn J L Ligtenberg, Cristina Martínez-Bouzas, Sonia Merino, Geneviève Michils, Srdjan Novaković, Ana Patiño-García, Guglielmina Nadia Ranzani, Evelin Schröck, Inês Silva, Catarina Silveira, José L Soto, Isabel Spier, Verena Steinke-Lange, Gianluca Tedaldi, María-Isabel Tejada, Emma R Woodward, Marc Tischkowitz, Noline Hoogerbrugge, Carla Oliveira

<https://pubmed.ncbi.nlm.nih.gov/36436516/>

Long-term chemoprevention in patients with adenomatous polyposis coli: an observational study.

Teresa M Neuhann, Katharina Haub, Verena Steinke-Lange, Monika Morak, Andreas Laner, Melanie Locher, Elke Holinski-Feder.

<https://pubmed.ncbi.nlm.nih.gov/35570229/>

Germline mutations in WNK2 could be associated with serrated polyposis syndrome.

Soares de Lima Y, Arnau-Collell C, Muñoz J, Herrera-Pariente C, Moreira L, Ocaña T, Díaz-Gay M, Franch-Expósito S, Cuatrecasas M, Carballal S, Lopez-Novo A, Moreno L, Fernández G, Díaz de Bustamante A, Peters S, Sommer AK, Spier I, Te Paske IBAW, van Herwaarden YJ, Castells A, Bujanda L, Capellà G, Steinke-Lange V, Mahmood K, Joo JE, Arnold J, Parry S, Macrae FA, Winship IM, Rosty C, Cubiella J, Rodríguez-Alcalde D, Holinski-Feder E, de Voer R, Buchanan DD, Aretz S, Ruiz-Ponte C, Valle L, Balaguer F, Bonjoch L, Castellvi-Bel S.

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Colorectal cancer incidences in Lynch syndrome: a comparison of results from the prospective lynch syndrome database and the international mismatch repair consortium.

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Constitutional chromothripsis of the APC locus as a cause of genetic predisposition to colon cancer

Florentine Scharf, Rafaela Magalhaes Leal Silva, Monika Morak, Alex Hastie, Julia M A Pickl, Kai Sendelbach, Christian Gebhard, Melanie Locher, Andreas Laner, Verena Steinke-Lange, Udo Koehler, Elke Holinski-Feder, Dieter A Wolf

<https://pubmed.ncbi.nlm.nih.gov/34911816/>

Cancer risks by sex and variant type in PTEN Hamartoma Tumor Syndrome.

Hendricks LAJ, Hoogerbrugge N, Mensenkamp AR, Brunet J, Lleuger-Pujol R, Høberg-Vetti H, Haavind MT, Innella G, Turchetti D, Aretz S, Spier I, Tischkowitz M, Jahn A, Links TP, Olderode-Berends MJW, Blatnik A, Leter EM, Evans DG, Woodward ER, Steinke-Lange V, Anastasiadou VC, Colas C, Villy MC, Benusiglio PR, Gerasimenko A, Barili V, Branchaud M, Houdayer C, Tesi B, Yazicioglu MO, van der Post RS, Schuurs-Hoeijmakers JHM; PTEN Study Group, Vos JR.

<https://pubmed.ncbi.nlm.nih.gov/36171661/>

Somatic copy number alteration and fragmentation analysis in circulating tumor DNA for cancer screening and treatment monitoring in colorectal cancer patients

Ariane Hallermayr, Tobias Wohlfrom, Verena Steinke-Lange, Anna Benet-Pagès, Florentine Scharf, Ellen Heitzer, Ulrich Mansmann, Christopher Haberl, Maike de Wit, Holger Vogelsang, Markus Rentsch, Elke Holinski-Feder, Julia M A Pickl

<https://pubmed.ncbi.nlm.nih.gov/36056434/>

Splicing analyses for variants in MMR genes: best practice recommendations from the European Mismatch Repair Working Group

Monika Morak, Marta Pineda, Alexandra Martins, Pascaline Gaildrat, Hélène Tubeuf, Aurélie Drouet, Carolina Gómez, Estela Dámaso, Kerstin Schaefer, Verena Steinke-Lange, Udo Koehler, Andreas Laner, Julie Hauchard, Karine Chauvis, Elke Holinski-Feder, Gabriel Capellà

<https://pubmed.ncbi.nlm.nih.gov/35676339/>

Long-term chemoprevention in patients with adenomatous polyposis coli: an observational study

Teresa M Neuhann, Katharina Haub, Verena Steinke-Lange, Monika Morak, Andreas Laner, Melanie Locher, Elke Holinski-Feder

<https://pubmed.ncbi.nlm.nih.gov/35570229/>

Solving the genetic aetiology of hereditary gastrointestinal tumour syndromes- a collaborative multicentre endeavour within the project Solve-RD

Anna K Sommer, Iris B A W Te Paske, José Garcia-Pelaez, Andreas Laner, Elke Holinski-Feder, Verena Steinke-Lange, Sophia Peters, Laura Valle, Isabel Spier, David Huntsman, Solve-RD-GENTURIS group; Carla Oliveira, Richarda M de Voer, Nicoline Hoogerbrugge, Stefan Aretz

<https://doi.org/10.1016/j.ejmg.2022.104475>

Clinical Validity of Circulating Tumor DNA as Prognostic and Predictive Marker for Personalized Colorectal Cancer Patient Management

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<https://doi.org/10.3390/cancers14030851>

Overview of the Clinical Features of Li-Fraumeni Syndrome and the Current European ERN GENTURIS Guideline

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<https://doi.org/10.1055/a-1541-7912>

2021

Constitutional chromothripsis of the APC locus as a cause of genetic predisposition to colon cancer

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<https://doi.org/10.1136/jmedgenet-2021-108147>

Somatic mosaics in hereditary tumor predisposition syndromes

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<https://doi.org/10.1016/j.ejmg.2021.104360>

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