

2026

The burden of TTN variants in the genomic era: Analysis of 18,462 individuals from the Solve-RD consortium and general recommendations.

Di Feo MF, Paramonov I, Matalonga Borrel L, Töpf A, Hoischen A, Beltran S, Graessner H, Vissers L, de Voer R, van Gijn M, Balestrini S, Lerche H, Lesca G, Gayathri SN, Ellwanger K, Cossee M, Perrin A, Sarkozy A, Bonne G, Verdonschot JAJ, Demidov G, Laurie S, Johari M; Solve-RD consortium (Steinke-Lange V et al.), Hackman P, Savarese M, Udd B.

<https://pubmed.ncbi.nlm.nih.gov/41277541/>

Effect of a polygenic risk score in patients with late-onset, early-onset, familial, or hereditary colorectal cancer.

Klinkhammer H, Spier I, Perne C, Hoffmann P, Büttner R, Schröck E, Redler S, Jahn A, Franke M, Möslin G, Kloor M, Schmidt B, Vangala D, Nguyen HP, Forstner AJ, Steinke-Lange V, Holinski-Feder E, Hüneburg R, Maj C, Engel C, Mayr A, Aretz S.

<https://pubmed.ncbi.nlm.nih.gov/41975491/>

Mutational Landscape of Colorectal Tumors From Individuals With Unexplained Adenomatous or Serrated Colorectal Polyposis.

Sommer AK, Te Paske IBAW, Jansen EAM, Gschwind A, Demidov G, Steinke-Lange V, Spier I, Steyaert W, Yaldiz B, Steehouwer M, Hommerding O, Dietrich D, Peters S, Garcia-Pelaez J, Laner A, Oliveira C, Capellá G, Nagtegaal ID, Bläker H, Ellwanger K, Hoischen A, Gilissen C, van der Post RS, Kristiansen G, Hoogerbrugge N, Ossowski S, Valle L, Holinski-Feder E, Ligtenberg MJL, Aretz S, de Voer RM.

<https://pubmed.ncbi.nlm.nih.gov/41524686/>

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.

<https://pubmed.ncbi.nlm.nih.gov/41347847/>

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/>

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.

<https://pubmed.ncbi.nlm.nih.gov/40468016/>

The risk of a second primary cancer in PTEN Hamartoma Tumor Syndrome (PHTS).

Hendricks LAJ, Verbeek KCJ, Schuurs-Hoeijmakers JHM, de Jong MM, Links TP, Brems H, Aerden M, Brunet J, Lleuger-Pujol R, Hüneburg R, Aretz S, Colas C, Villy MC, Woodward ER, Evans DG, Bosch DGM, Donze SH, Foretová L, Blatnik A, Leter EM, Tischkowitz M, Jahn A, de Putter R, Dupont J, Briskemyr S, Steinke-Lange V, Baldassarri M,

Anastasiadou VC, Irmejs A, Oliveira C, van der Post RS, Mensenkamp AR, Tesi B, Mu N, Benusiglio PR, Gerasimenko A, Innella G, Turchetti D, Houdayer C, Branchaud M, Høberg-Vetti H, Haavind MT, Balmaña J, Torres M, Genuardi M, Panfili A, Jørgensen K, Mæhle L, Hoogerbrugge N, Vos JR.

<https://pubmed.ncbi.nlm.nih.gov/40433764/>

Colonoscopy findings in CDH1 carriers from a multicenter international study.

Chatterjee A, Hüneburg R, Yang Q, Morrison S, Bettzüge A, Marwitz T, Aretz S, Spier I, Ripperger T, Redler S, Kachanov M, Volk AE, Vangala DB, Daum S, Holinski-Feder E, Steinke-Lange V, Bahlke K, Strassburg CP, MejiaPerez LK, O'Malley MM, LaGuardia L, Liska D, Macaron C, Sommovilla J, Burke CA, Nattermann J.

<https://pubmed.ncbi.nlm.nih.gov/40323501/>

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

Lucas MC, Keßler T, Scharf F, Steinke-Lange V, Klink B, Laner A, Holinski-Feder E.

<https://pubmed.ncbi.nlm.nih.gov/40317406/>

Joint analysis of germline genetic data from over 29,000 cases with suspected hereditary breast and ovarian cancer (HBOC) as part of the NASGE initiative.

Henkel J, Laner A, Locher M, Wohlfrom T, Neitzel B, Becker K, Neuhann T, Abicht A, Steinke-Lange V, Klink B, Eichhorn B, Schmidt W, Berner D, Teubert A, Holtorf A, Heinrich S, Wildhardt G, Schulze M, von der Heyden L, Hörtnagel K, Steinberger D, Kleier S, Lorenz P, Glaubitz R, Biskup S, Holinski-Feder E.

<https://pubmed.ncbi.nlm.nih.gov/39854808/>

Metachronous colorectal cancer risks after extended or segmental resection in MLH1, MSH2, and MSH6 Lynch syndrome: multicentre study from the Prospective Lynch Syndrome Database.

Prospective Lynch Syndrome Database (u. a. Holinski-Feder E, Steinke-Lange V et al.).

<https://pubmed.ncbi.nlm.nih.gov/40231433/>

Genomic reanalysis of a pan-European rare-disease resource yields new diagnoses.

Laurie S, Steyaert W, de Boer E, Polavarapu K, Schuermans N, Sommer AK, Demidov G, Ellwanger K, Paramonov I, Thomas C, Aretz S, Baets J, Benetti E, Bullich G, Chinnery PF, Clayton-Smith J, Cohen E, Danis D, de Sainte Agathe JM, Denommé-Pichon AS, Diaz-Manera J, Efthymiou S, Faivre L, Fernandez-Callejo M, Freeberg M, Garcia-Pelaez J, Guillot-Noel L, Haack TB, Hanna M, Hengel H, Horvath R, Houlden H, Jackson A, Johansson L, Johari M, Kamsteeg EJ, Kellner M, Kleefstra T, Lacombe D, Lochmüller H, López-Martín E, Macaya A, Marcé-Grau A, Maver A, Morsy H, Muntoni F, Musacchia F, Nelson I, Nigro V, Olimpio C, Oliveira C, Paulasová Schwabová J, Pauly MG, Peterlin B, Peters S, Pfundt R, Piluso G, Piscia D, Posada M, Reich S, Renieri A, Ryba L, Šablauskas K, Savarese M, Schöls L, Schütz L, Steinke-Lange V, Stevanin G, Straub V, Sturm M, Swertz MA, Tartaglia M, Te Paske IBAW, Thompson R, Torella A, Trainor C, Udd B, Van de Vondel L, van de Warrenburg B, van Reeuwijk J, Vandrovцова J, Vitobello A, Vos J, Vyhnáková E, Wijngaard R, Wilke C, William D, Xu J, Yaldiz B, Zalatnai L, Zurek B, Solve-RD DITF-GENTURIS, Solve-RD DITF-ITHACA, Solve-RD DITF-EURO-NMD, Solve-RD DITF-RND, Solve-RD consortium, Brookes AJ, Evangelista T, Gilissen C, Graessner H, Hoogerbrugge N, Ossowski S, Riess O, Schüle R, Synofzik M, Verloes A, Matalonga L, Brunner HG, Lohmann K, de Voer RM, Töpf A, Vissers LELM, Beltran S, Hoischen A.

<https://pubmed.ncbi.nlm.nih.gov/39825153/>

Reclassification of VUS in BRCA1 and BRCA2 using the new BRCA1/BRCA2 ENIGMA track set demonstrates the superiority of ClinGen ENIGMA Expert Panel specifications over the standard ACMG/AMP classification system.

Benet-Pagès A, Laner A, Nassar LR, Wohlfrom T, Steinke-Lange V, Haeussler M, Holinski-Feder E.

<https://pubmed.ncbi.nlm.nih.gov/40027238/>

2024

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

Biermann S, Knapp M, Wieacker P, Aretz S, Steinke-Lange V.

<https://pubmed.ncbi.nlm.nih.gov/38280980/>

Comprehensive reanalysis for CNVs in ES data from unsolved rare disease cases results in new diagnoses.

Demidov G, Yaldiz B, Garcia-Pelaez J, de Boer E, Schuermans N, Van de Vondel L, Paramonov I, Johansson LF, Musacchia F, Benetti E, Bullich G, Sablauskas K, Beltran S, Gilissen C, Hoischen A, Ossowski S, de Voer R, Lohmann K, Oliveira C, Topf A, Vissers LELM, Solve-RD Consortium (Steinke-Lange et al.), Laurie S.

<https://pubmed.ncbi.nlm.nih.gov/39461972/>

[Lynch syndrome].

Steinke-Lange V, Holinski-Feder E.

<https://pubmed.ncbi.nlm.nih.gov/38864870/>

Cancer predisposing syndromes in childhood and adolescence pose several challenges necessitating interdisciplinary care in dedicated programs.

Kaffai S, Angelova-Toshkin D, Weins AB, Ickinger S, Steinke-Lange V, Vollert K, Frühwald MC, Kuhlen M.

<https://pubmed.ncbi.nlm.nih.gov/38887560/>

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

Hendricks LAJ, Verbeek KCJ, Schuurs-Hoeijmakers JHM, Mensenkamp AR, Brems H, de Putter R, Anastasiadou VC, Villy MC, Jahn A, Steinke-Lange V, Baldassarri M, Irmejs A, de Jong MM, Links TP, Leter EM, Bosch DGM, Høberg-Vetti H, Tveit Haavind M, Jørgensen K, Mæhle L, Blatnik A, Brunet J, Darder E, Tham E, Hoogerbrugge N, Vos JR.

<https://pubmed.ncbi.nlm.nih.gov/38473316/>

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

Johansson LF, Laurie S, Spalding D, Gibson S, Ruvoilo D, Thomas C, Piscia D, de Andrade F, Been G, Bijlsma M, Brunner H, Cimerman S, Dizjikan FY, Ellwanger K, Fernandez M, Freeberg M, van de Geijn GJ, Kanninga R, Maddi V, Mehtarizadeh M, Neerinx P, Ossowski S, Rath A, Roelofs-Prins D, Stok-Benjamins M, van der Velde KJ, Veal C, van der Vries G, Wadsley M, Warren G, Zurek B, Keane T, Graessner H, Beltran S, Swertz MA, Brookes AJ, Solve-RD consortium (Steinke-Lange V et al.).

<https://pubmed.ncbi.nlm.nih.gov/39302238/>

2023

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

Hallermayr A, Keßler T, Steinke-Lange V, Heitzer E, Holinski-Feder E, Speicher M.

<https://pubmed.ncbi.nlm.nih.gov/38835734/>

Impact of cfDNA Reference Materials on Clinical Performance of Liquid Biopsy NGS Assays.

Hallermayr A, Keßler T, Fujera M, Liesfeld B, Bernstein S, von Ameln S, Schanze D, Steinke-Lange V, Pickl JMA, Neuhann TM, Holinski-Feder E.

<https://pubmed.ncbi.nlm.nih.gov/37894392/>

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

São José C, Garcia-Pelaez J, Ferreira M, Arrieta O, André A, Martins N, Solís S, Martínez-Benítez B, Ordóñez-Sánchez ML, Rodríguez-Torres M, Sommer AK, Te Paske IBAW, Caldas C, Tischkowitz M, Tusié MT, Solve-RD DITF-GENTURIS (Steinke-Lange V et al.), Hoogerbrugge N, Demidov G, de Voer RM, Laurie S, Oliveira C.

<https://pubmed.ncbi.nlm.nih.gov/37249750/>

Endometrial Cancer. Guideline of the DGGG, DKG and DKH (S3-Level, AWMF Registry Number 032/034-OL, September 2022). Part 1 with Recommendations on the Epidemiology, Screening, Diagnosis and Hereditary Factors of Endometrial Cancer, Geriatric Assessment and Supply Structures.

Emons G, Steiner E, Vordermark D, Uleer C, Paradies K, Tempfer C, Aretz S, Cremer W, Hanf V, Mallmann P, Ortmann O, Römer T, Schmutzler RK, Horn LC, Kommos S, Lax S, Schmoeckel E, Mokry T, Grab D, Reinhardt M, Steinke-Lange V, Brucker SY, Kiesel L, Witteler R, Fleisch MC; Heinrich Prömpeler † 25; Friedrich M, Höcht S, Lichtenegger W, Mueller M, Runnebaum I, Feyer P, Hagen V, Juhasz-Böss I, Letsch A, Niehoff P, Zeimet AG, Battista MJ, Petru E, Widhalm S, van Oorschot B, Panke JE, Weis J, Dauelsberg T, Haase H, Beckmann MW, Jud S, Wight E, Prott FJ, Micke O, Bader W, Reents N, Henschler U; Reina Tholen † 52; Schallenberg M, Rahner N, Mayr D, Kreißl M, Lindel K, Mustea A, Strnad V, Goerling U, Bauerschmitz GJ, Langrehr J, Neulen J, Ulrich UA, Nothacker MJ, Blödt S, Follmann M, Langer T, Wenzel G, Weber S, Erdogan S.

<https://pubmed.ncbi.nlm.nih.gov/37588260/>

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.

<https://pubmed.ncbi.nlm.nih.gov/37396538/>

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.

<https://pubmed.ncbi.nlm.nih.gov/37188824/>

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 Heinimann, 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 Schmiegel, 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.

<https://pubmed.ncbi.nlm.nih.gov/37181409/>

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

Hallermayr A, Neuhann TM, Steinke-Lange V, Scharf F, Laner A, Ewald R, Liesfeld B, Holinski-Feder E, Pickl JMA.

<https://pubmed.ncbi.nlm.nih.gov/36636551/>

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

<https://pubmed.ncbi.nlm.nih.gov/36593122/>

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 García-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.

<https://pubmed.ncbi.nlm.nih.gov/36270769/>

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

Møller P, Seppälä T, Dowty JG, Haupt S, Dominguez-Valentin M, Sunde L, Bernstein I, Engel C, Aretz S, Nielsen M, Capella G, Evans DG, Burn J, Holinski-Feder E, Bertario L, Bonanni B, Lindblom A, Levi Z, Macrae F, Winship I, Plazzer JP, Sijmons R, Laghi L, Valle AD, Heinimann K, Half E, Lopez-Koestner F, Alvarez-Valenzuela K, Scott RJ, Katz L, Laish I, Vainer E, Vaccaro CA, Carraro DM, Gluck N, Abu-Freha N, Stakelum A, Kennelly R, Winter D, Rossi BM, Greenblatt M, Bohorquez M, Sheth H, Tibiletti MG, Lino-Silva LS, Horisberger K, Portenkirchner C, Nascimento I, Rossi NT, da Silva LA, Thomas H, Zaránd A, Mecklin JP, Pylvänäinen K, Renkonen-Sinisalo L, Lepisto A, Peltomäki P, Therkildsen C, Lindberg LJ, Thorlacius-Ussing O, von Knebel Doeberitz M, Loeffler M, Rahner N, Steinke-Lange V, Schmiegel W, Vangala D, Perne C, Hüneburg R, de Vargas AF, Latchford A, Gerdes AM, Backman AS, Guillén-Ponce C, Snyder C, Lautrup CK, Amor D, Palmero E, Stoffel E, Duijkers F, Hall MJ, Hampel H, Williams H, Okkels H, Lubiński J, Reece J, Ngeow J, Guillem JG, Arnold J, Wadt K, Monahan K, Senter L, Rasmussen LJ, van Hest LP, Ricciardiello L, Kohonen-Corish MRJ, Ligtenberg MJL, Southey M, Aronson M, Zahary MN, Samadder NJ, Poplawski N, Hoogerbrugge N, Morrison PJ, James P, Lee G, Chen-Shtoyerman R, Ankathil R, Pai R, Ward R, Parry S, Dębniak T, John T, van Overeem Hansen T, Caldés T, Yamaguchi T, Barca-Tierno V, Garre P, Cavestro GM, Weitz J, Redler S, Büttner R, Heuveline V, Hopper JL, Win AK, Lindor N, Gallinger S, Le Marchand L, Newcomb PA, Figueiredo J, Buchanan DD, Thibodeau SN, Ten Broeke SW, Hovig E, Nakken S, Pineda M, Dueñas N, Brunet J, Green K, Lalloo F, Newton K, Crosbie EJ, Mints M, Tjandra D, Neffa F, Esperon P, Kariv R, Rosner G, Pavicic WH, Kalfayan P, Torrezan GT, Basaneze T, Martin C, Moslein G, Ahadova A, Kloor M, Sampson JR, Jenkins MA; European Hereditary Tumour Group (EHTG) and the International Mismatch Repair Consortium (IMRC).

<https://pubmed.ncbi.nlm.nih.gov/36182917/>

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, Brancaud 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, Maïke 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, H el ene Tubeuf, Aur elie Drouet, Carolina G omez, Estela D amaso, Kerstin Schaefer, Verena Steinke-Lange, Udo Koehler, Andreas Laner, Julie Hauchard, Karine Chauaris, Elke Holinski-Feder, Gabriel Capell a

<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, Jos e 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

Ariane Hallermayr, Verena Steinke-Lange, Holger Vogelsang, Markus Rentsch, Maïke de Wit, Christopher Haberl, Elke Holinski-Feder, Julia M A Pickl

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

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

Christian Peter Kratz, Verena Steinke-Lange, Isabel Spier, Stefan Aretz, Evelin Schr ock, Elke Holinski-Feder

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

2021

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

Somatic mosaics in hereditary tumor predisposition syndromes

Verena Steinke-Lange, Robin de Putter, Elke Holinski-Feder, Kathleen Bm Claes

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

Adenoma and colorectal cancer risks in Lynch syndrome, Lynch-like syndrome, and familial colorectal cancer type X.

Bucksch K, Zachariae S, Ahadova A, Aretz S, B uttner R, G orgens H, Holinski-Feder E, H uneburg R, Kloor M, von Knebel Doeberitz M, Ladigan-Badura S, Moeslein G, Morak M, Nattermann J, Nguyen HP, Perne C, Redler S, Schmetz A, Steinke-Lange V, Surowy H, Vangala DB, Weitz J, Loeffler M, Engel C;

German Consortium for Familial Intestinal Cancer. Int J Cancer. 2021 Sep 1. <https://doi.org/10.1002/ijc.33790>

Liquid Biopsy Hotspot Variant Assays: Analytical Validation for Application in Residual Disease Detection and Treatment Monitoring.

Hallermayr A, Benet-Pag es A, Steinke-Lange V, Mansmann U, Rentsch M, Holinski-Feder E, Pickl JMA.

Clin Chem. 2021 Aug 15;hvb124, <https://doi.org/10.1093/clinchem/hvb124>

Early detection of duodenal cancer by upper GI-endoscopy in Lynch syndrome.

Vangala DB, Ladigan-Badura S, Engel C, Hüneburg R, Perne C, Bucksch K, Nattermann J, Steinke-Lange V, Rahner N, Weitz J, Kloor M, Tomann J, Canbay A, Nguyen HP, Strassburg C, Möslein G, Morak M, Holinski-Feder E, Büttner R, Aretz S, Loeffler M, Schmiegel W, Pox C, Schulmann K;
German Consortium for Familial Intestinal Cancer. *Int J Cancer*. 2021 Jul 31. <https://doi.org/10.1002/ijc.33753>

No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in MLH1 and MSH2: A Prospective Lynch Syndrome Database Study.

M. Dominguez-Valentin, J.-P. Plazzer, J. R. Sampson, C. Engel, S. Aretz, M. A. Jenkins, L. Sunde, I. Bernstein, G. Capella, F. Balaguer, F. Macrae, I. M. Winship, H. Thomas, D. G. Evans, J. Burn, M. Greenblatt, W. H. de Vos Tot Nederveen Cappel, R. H. Sijmons, M. Nielsen, L. Bertario, B. Bonanni, M. G. Tibiletti, G. M. Cavestro, A. Lindblom, A. D. Valle, F. Lopez-Kostner, K. Alvarez, N. Gluck, L. Katz, K. Heinimann, C. A. Vaccaro, S. Nakken, E. Hovig, K. Green, F. Laloo, J. Hill, H. F. A. Vasen, C. Perne, R. Büttner, H. Görgens, E. Holinski-Feder, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, N. Rahner, J. Weitz, **V. Steinke-Lange**, W. Schmiegel, D. Vangala, E. J. Crosbie, M. Pineda, M. Navarro, J. Brunet, L. Moreira, A. Sánchez, M. Serra-Burriel, M. Mints, R. Kariv, G. Rosner, T. A. Piñero, W. H. Pavicic, P. Kalfayan, S. W. T. Broeke, J.-P. Mecklin, K. Pylvänäinen, L. Renkonen-Sinisalo, A. Lepistö, P. Peltomäki, J. L. Hopper, A. K. Win, D. D. Buchanan, N. M. Lindor, S. Gallinger, L. L. Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, C. Therkildsen, T. V. O. Hansen, L. Lindberg, E. A. Rødland, F. Neffa, P. Esperon, D. Tjandra, G. Möslein, T. T. Seppälä, & P. Møller, *Journal of Clinical Medicine*, 10 (2021) 2856. <https://doi.org/10.3390/jcm10132856>.

Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases.

B. Zurek, K. Ellwanger, L. E. L. M. Vissers, R. Schüle, M. Synofzik, A. Töpf, R. M. de Voer, S. Laurie, L. Matalonga, C. Gilissen, S. Ossowski, P. A. C. 't Hoen, A. Vitobello, J. M. Schulze-Hentrich, O. Riess, H. G. Brunner, A. J. Brookes, A. Rath, G. Bonne, G. Gumus, A. Verloes, N. Hoogerbrugge, T. Evangelista, T. Harmuth, M. Swertz, D. Spalding, A. Hoischen, S. Beltran, H. Graessner, & Solve-RD consortium, *European journal of human genetics: EJHG*, (2021). <https://doi.org/10.1038/s41431-021-00859-0>.

Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study.

International Mismatch Repair Consortium. *The Lancet. Oncology*, (2021) S1470-2045(21)00189-3. [https://doi.org/10.1016/S1470-2045\(21\)00189-3](https://doi.org/10.1016/S1470-2045(21)00189-3)

Hereditary non-polyposis tumor risk syndromes

V. Steinke-Lange & E. Holinski-Feder. *MMW Fortschritte der Medizin*, 163 (2021) 41-44. <https://doi.org/10.1007/s15006-021-9960-1>

A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. I. B. A. W. Te Paske, J. Garcia-Pelaez, A. K. Sommer, L. Matalonga, T. Starzynska, A. Jakubowska, Solve-RD-GENTURIS group, R. S. van der Post, J. Lubinski, C. Oliveira, N. Hoogerbrugge, & R. M. de Voer, *European journal of human genetics: EJHG*, (2021). <https://doi.org/10.1038/s41431-021-00853-6>

Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. E. Legius, L. Messiaen, P. Wolkenstein, P. Pancza, R. A. Avery, Y. Berman, J. Blakeley, D. Babovic-Vuksanovic, K. S. Cunha, R. Ferner, M. J. Fisher, J. M. Friedman, D. H. Gutmann, H. Kehrer-Sawatzki, B. R. Korf, V.-F. Mautner, S. Peltonen, K. A. Rauen, V. Riccardi, E. Schorry, A. Stemmer-Rachamimov, D. A. Stevenson, G. Tadini, N. J. Ullrich, D. Viskochil, K. Wimmer, K. Yohay, International Consensus Group on Neurofibromatosis Diagnostic Criteria (I-NF-DC), S. M. Huson, D. G. Evans, & S. R. Plotkin, *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, (2021). <https://doi.org/10.1038/s41436-021-01170-5>

Uptake of hysterectomy and bilateral salpingo-oophorectomy in carriers of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report.

T. T. Seppälä, M. Dominguez-Valentin, E. J. Crosbie, C. Engel, S. Aretz, F. Macrae, I. Winship, G. Capella, H. Thomas, E. Hovig, M. Nielsen, R. H. Sijmons, L. Bertario, B. Bonanni, M. G. Tibiletti, G. M. Cavestro, M. Mints, N. Gluck, L. Katz, K. Heinimann, C. A. Vaccaro, K. Green, F. Laloo, J. Hill, W. Schmiegel, D. Vangala, C. Perne, H.-G. Strauß, J. Tecklenburg, E. Holinski-Feder, V. Steinke-Lange, J.-P. Mecklin, J.-P. Plazzer, M. Pineda, M. Navarro, J. B. Vida, R. Kariv, G. Rosner, T. A. Piñero, W. Pavicic, P. Kalfayan, S. W. Ten Broeke, M. A. Jenkins, L. Sunde, I. Bernstein, J. Burn, M. Greenblatt, W. H. de Vos Tot Nederveen Cappel, A. Della Valle, F. Lopez-Koestner, K. Alvarez, R. Büttner, H. Görgens,

M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, S. Redler, J. Weitz, K. Pylvänäinen, L. Renkonen-Sinisalo, A. Lepistö, J. L. Hopper, A. K. Win, N. M. Lindor, S. Gallinger, L. Le Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, C. Therkildsen, K. A. W. Wadt, M. J. E. Mourits, Z. Ketabi, O. G. Denton, E. A. Rødland, H. Vasen, F. Neffa, P. Esperon, D. Tjandra, G. Möslin, E. Rökkones, J. R. Sampson, D. G. Evans, & P. Møller, *European Journal of Cancer* (Oxford, England: 1990), 148 (2021) 124–133. <https://doi.org/10.1016/j.ejca.2021.02.022>

Risk-reducing hysterectomy and bilateral salpingo-oophorectomy in female heterozygotes of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report.

M. Dominguez-Valentin, E. J. Crosbie, C. Engel, S. Aretz, F. Macrae, I. Winship, G. Capella, H. Thomas, S. Nakken, E. Hovig, M. Nielsen, R. H. Sijmons, L. Bertario, B. Bonanni, M. G. Tibiletti, G. M. Cavestro, M. Mints, N. Gluck, L. Katz, K. Heinimann, C. A. Vaccaro, K. Green, F. Laloo, J. Hill, W. Schmiegell, D. Vangala, C. Perne, H.-G. Strauß, J. Tecklenburg, E. Holinski-Feder, V. Steinke-Lange, J.-P. Mecklin, J.-P. Plazzer, M. Pineda, M. Navarro, J. B. Vidal, R. Kariv, G. Rosner, T. A. Piñero, M. L. Gonzalez, P. Kalfayan, N. Ryan, S. W. Ten Broeke, M. A. Jenkins, L. Sunde, I. Bernstein, J. Burn, M. Greenblatt, W. H. de Vos Tot Nederveen Cappel, A. Della Valle, F. Lopez-Koestner, K. Alvarez, R. Büttner, H. Görgens, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, N. Rahner, J. Weitz, K. Pylvänäinen, L. Renkonen-Sinisalo, A. Lepistö, A. Auranen, J. L. Hopper, A. K. Win, R. W. Haile, N. M. Lindor, S. Gallinger, L. Le Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, C. Therkildsen, H. Okkels, Z. Ketabi, O. G. Denton, E. A. Rødland, H. Vasen, F. Neffa, P. Esperon, D. Tjandra, G. Möslin, J. R. Sampson, D. G. Evans, T. T. Seppälä, & P. Møller, *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, 23 (2021) 705–712. <https://doi.org/10.1038/s41436-020-01029-1>.

Value of upper gastrointestinal endoscopy for gastric cancer surveillance in patients with Lynch syndrome. S. Ladigan-Badura, D. B. Vangala, C. Engel, K. Bucksch, R. Hueneburg, C. Perne, J. Nattermann, V. Steinke-Lange, N. Rahner, H. K. Schackert, J. Weitz, M. Kloor, J. Kuhlkamp, H. P. Nguyen, G. Moeslein, C. Strassburg, M. Morak, E. Holinski-Feder, R. Buettner, S. Aretz, M. Loeffler, W. Schmiegell, C. Pox, K. Schulmann, & German Consortium for Familial Intestinal Cancer, *International Journal of Cancer*, 148 (2021) 106–114. <https://doi.org/10.1002/ijc.33294>

2020

Age-dependent performance of BRAF mutation testing in Lynch syndrome diagnostics. Bläker H, Haupt S, Morak M, Holinski-Feder E, Arnold A, Horst D, Sieber-Frank J, Seidler F, von Winterfeld M, Alwers E, Chang-Claude J, Brenner H, Roth W, Engel C, Löffler M, Möslin G, Schackert H-K, Weitz J, Perne C, Aretz S, Hüneburg R, Schmiegell W, Vangala D, Rahner N, Steinke-Lange V, Heuveline V, von Knebel Doeberitz M, Ahadova A, Hoffmeister M, Kloor M. *Int J Cancer*. 2020;147(10):2801-2810. doi:[10.1002/ijc.33273](https://doi.org/10.1002/ijc.33273)

Value of upper GI endoscopy for gastric cancer surveillance in patients with Lynch syndrome.

Ladigan-Badura S, Vangala DB, Engel C, Bucksch K, Hueneburg R, Perne C, Nattermann J, Steinke-Lange V, Rahner N, Schackert HK, Weitz J, Kloor M, Kuhlkamp J, Nguyen HP, Moeslein G, Strassburg C, Morak M, Holinski-Feder E, Buettner R, Aretz S, Loeffler M, Schmiegell W, Pox C, Schulmann K. *Int J Cancer*. Published online September 15, 2020. doi:[10.1002/ijc.33294](https://doi.org/10.1002/ijc.33294)

Rare tumors as leading symptom of hereditary tumor syndromes.

Perne C, Steinke-Lange V, Aretz S, Spier I. *Pathologe*. 2020;41(5):535-549. doi:[10.1007/s00292-020-00806-8](https://doi.org/10.1007/s00292-020-00806-8)

The “unnatural” history of colorectal cancer in Lynch syndrome: Lessons from colonoscopy surveillance.

Ahadova A, Seppälä TT, Engel C, Gallon R, Burn J, Holinski-Feder E, Steinke-Lange V, Möslin G, Nielsen M, Ten Broeke SW, Laghi L, Dominguez-Valentin M, Capella G, Macrae F, Scott R, Hüneburg R, Nattermann J, Hoffmeister M, Brenner H, Bläker H, von Knebel Doeberitz M, Sampson JR, Vasen H, Mecklin J-P, Møller P, Kloor M. *Int J Cancer*. Published online July 19, 2020. doi:[10.1002/ijc.33224](https://doi.org/10.1002/ijc.33224)

Risk-Reducing Gynecological Surgery in Lynch Syndrome: Results of an International Survey from the Prospective Lynch Syndrome Database.

Dominguez-Valentin M, Seppälä TT, Engel C, Aretz S, Macrae F, Winship I, Capella G, Thomas H, Hovig E, Nielsen M, Sijmons RH, Bertario L, Bonanni B, Tibiletti MG, Cavestro GM, Mints M, Gluck N, Katz L, Heinimann K, Vaccaro CA, Green K, Laloo F, Hill J, Schmiegell W, Vangala D, Perne C, Strauß H-G, Tecklenburg J, Holinski-Feder E, Steinke-Lange V, Mecklin J-P, Plazzer J-P, Pineda M, Navarro M, Vidal JB, Kariv R, Rosner G, Piñero TA, Gonzalez ML, Kalfayan P, Sampson JR, Ryan N AJ, Evans DG, Møller P, Crosbie EJ. *J Clin Med*. 2020;9(7). doi:[10.3390/jcm9072290](https://doi.org/10.3390/jcm9072290)

Cancer risks in Lynch syndrome, Lynch-like syndrome, and familial colorectal cancer type X: a prospective cohort study. Bucksch K, Zachariae S, Aretz S, Büttner R, Holinski-Feder E, Holzapfel S, Hüneburg R, Kloor M, von Knebel Doeberitz M, Morak M, Möslin G, Nattermann J, Perne C, Rahner N, Schmiegel W, Schulmann K, Steinke-Lange V, Strassburg CP, Vangala DB, Weitz J, Loeffler M, Engel C.
BMC Cancer. 2020;20(1):460. doi:[10.1186/s12885-020-06926-x](https://doi.org/10.1186/s12885-020-06926-x)

Prevalence of CNV-neutral structural genomic rearrangements in MLH1, MSH2, and PMS2 not detectable in routine NGS diagnostics.
Morak M, Steinke-Lange V, Massdorf T, Benet-Pages A, Locher M, Laner A, Kayser K, Aretz S, Holinski-Feder E.
Fam Cancer. 2020;19(2):161-167. doi:[10.1007/s10689-020-00159-4](https://doi.org/10.1007/s10689-020-00159-4)

Associations of Pathogenic Variants in MLH1, MSH2, and MSH6 With Risk of Colorectal Adenomas and Tumors and With Somatic Mutations in Patients With Lynch Syndrome.
Engel C, Ahadova A, Seppälä TT, Aretz S, Bigirwamungu-Bargeman M, Bläker H, Bucksch K, Büttner R, de Vos Tot Nederveen Cappel WT, Endris V, Holinski-Feder E, Holzapfel S, Hüneburg R, Jacobs MAJM, Koornstra JJ, Langers AM, Lepistö A, Morak M, Möslin G, Peltomäki P, Pylvänäinen K, Rahner N, Renkonen-Sinisalo L, Schulmann K, **Steinke-Lange V**, Stenzinger A, Strassburg CP, van de Meeberg PC, van Kouwen M, van Leerdam M, Vangala DB, Vecht J, Verhulst M-L, von Knebel Doeberitz M, Weitz J, Zachariae S, Loeffler M, Mecklin J-P, Kloor M, Vasen HF.
Gastroenterology. 2020;158(5):1326-1333. doi:[10.1053/j.gastro.2019.12.032](https://doi.org/10.1053/j.gastro.2019.12.032)

Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database.
Dominguez-Valentin M, Sampson JR, Seppälä TT, Ten Broeke SW, Plazzer J-P, Nakken S, Engel C, Aretz S, Jenkins MA, Sunde L, Bernstein I, Capella G, Balaguer F, Thomas H, Evans DG, Burn J, Greenblatt M, Hovig E, de Vos Tot Nederveen Cappel WH, Sijmons RH, Bertario L, Tibiletti MG, Cavestro GM, Lindblom A, Della Valle A, Lopez-Köstner F, Gluck N, Katz LH, Heinimann K, Vaccaro CA, Büttner R, Görgens H, Holinski-Feder E, Morak M, Holzapfel S, Hüneburg R, Knebel Doeberitz M von, Loeffler M, Rahner N, Schackert HK, Steinke-Lange V, Schmiegel W, Vangala D, Pylvänäinen K, Renkonen-Sinisalo L, Hopper JL, Win AK, Haile RW, Lindor NM, Gallinger S, Le Marchand L, Newcomb PA, Figueiredo JC, Thibodeau SN, Wadt K, Therikildsen C, Okkels H, Ketabi Z, Moreira L, Sánchez A, Serra-Burriel M, Pineda M, Navarro M, Blanco I, Green K, Laloo F, Crosbie EJ, Hill J, Denton OG, Frayling IM, Rødland EA, Vasen H, Mints M, Neffa F, Esperon P, Alvarez K, Kariv R, Rosner G, Pinero TA, Gonzalez ML, Kalfayan P, Tjandra D, Winship IM, Macrae F, Möslin G, Mecklin J-P, Nielsen M, Møller P.
Genet Med. 2020;22(1):15-25. doi:[10.1038/s41436-019-0596-9](https://doi.org/10.1038/s41436-019-0596-9)

2019

Lack of association between screening interval and cancer stage in Lynch syndrome may be accounted for by over-diagnosis; a prospective Lynch syndrome database report. Seppälä TT, Ahadova A, Dominguez-Valentin M, Macrae F, Evans DG, Therikildsen C, Sampson J, Scott R, Burn J, Möslin G, Bernstein I, Holinski-Feder E, Pylvänäinen K, Renkonen-Sinisalo L, Lepistö A, Lautrup CK, Lindblom A, Plazzer J-P, Winship I, Tjandra D, Katz LH, Aretz S, Hüneburg R, Holzapfel S, Heinimann K, Valle AD, Neffa F, Gluck N, de Vos tot Nederveen Cappel WH, Vasen H, Morak M, Steinke-Lange V, Engel C, Rahner N, Schmiegel W, Vangala D, Thomas H, Green K, Laloo F, Crosbie EJ, Hill J, Capella G, Pineda M, Navarro M, Blanco I, ten Broeke S, Nielsen M, Ljungmann K, Nakken S, Lindor N, Frayling I, Hovig E, Sunde L, Kloor M, Mecklin J-P, Kalager M, Møller P.
Hered Cancer Clin Pract. 2019;17(1):8. doi:[10.1186/s13053-019-0106-8](https://doi.org/10.1186/s13053-019-0106-8)

Full-length transcript amplification and sequencing as universal method to test mRNA integrity and biallelic expression in mismatch repair genes.
Morak M, Schaefer K, Steinke-Lange V, Koehler U, Keinath S, Massdorf T, Mauracher B, Rahner N, Bailey J, Kling C, Haeusser T, Laner A, Holinski-Feder E. *Eur J Hum Genet.* 2019;27(12):1808-1820. doi:[10.1038/s41431-019-0472-8](https://doi.org/10.1038/s41431-019-0472-8)

Current recommendations for surveillance, risk reduction and therapy in Lynch syndrome patients.
Hüneburg R, Aretz S, Büttner R, Daum S, Engel C, Fechner G, Habermann JK, Heling D, Hoffmann K, Holinski-Feder E, Kloor M, von Knebel-Döberitz M, Loeffler M, Möslin G, Perne C, Redler S, Rieß O, Schmiegel W, Seufferlein T, Siebers-Renelt U, Steinke-Lange V, Tecklenburg J, Vangala D, Vilz T, Weitz J, Wiedenmann B, Strassburg CP, Nattermann J. *Z Gastroenterol.* 2019;57(11):1309-1320. doi:[10.1055/a-1008-9827](https://doi.org/10.1055/a-1008-9827)

Genetic Screening and Personalized Prevention in Colorectal Cancer.

Steinke-Lange V, Holinski-Feder E. *Visc Med.* 2019;35(4):226-230. doi:[10.1159/000501941](https://doi.org/10.1159/000501941)

The Apparent Genetic Anticipation in PMS2-Associated Lynch Syndrome Families Is Explained by Birth-cohort Effect.

Ten Broeke SW, Rodríguez-Girondo M, Suerink M, Aretz S, Bernstein I, Capellá G, Engel C, Gomez-Garcia EB, van Hest LP, von Knebel Doeberitz M, Lagerstedt-Robinson K, Letteboer TGW, Moller P, van Os TA, Pineda M, Rahner N, Olderoode-Berends MJW, von Salomé J, Schackert HK, Spruijt L, Steinke-Lange V, Wagner A, Tops CMJ, Nielsen M. *Cancer Epidemiol Biomarkers Prev.* 2019;28(6):1010-1014. doi:[10.1158/1055-9965.EPI-18-0576](https://doi.org/10.1158/1055-9965.EPI-18-0576)

Survival by colon cancer stage and screening interval in Lynch syndrome: a prospective Lynch syndrome database report.

Dominguez-Valentin M, Seppälä TT, Sampson JR, Macrae F, Winship I, Evans DG, Scott RJ, Burn J, Möslein G, Bernstein I, Pylvänäinen K, Renkonen-Sinisalo L, Lepistö A, Lindblom A, Plazzer J-P, Tjandra D, Thomas H, Green K, Laloo F, Crosbie EJ, Hill J, Capella G, Pineda M, Navarro M, Vidal JB, Rønlund K, Nielsen RT, Yilmaz M, Elvang LL, Katz L, Nielsen M, Ten Broeke SW, Nakken S, Hovig E, Sunde L, Kloor M, Knebel Doeberitz MV, Ahadova A, Lindor N, Steinke-Lange V, Holinski-Feder E, Mecklin J-P, Møller P.

Hered Cancer Clin Pract. 2019;17:28. doi:[10.1186/s13053-019-0127-3](https://doi.org/10.1186/s13053-019-0127-3)

2018

Copy number variation analysis and targeted NGS in 77 families with suspected Lynch syndrome reveals novel potential causative genes.

Kayser K, Degenhardt F, Holzapfel S, Horpaopan S, Peters S, Spier I, Morak M, Vangala D, Rahner N, von Knebel-Doeberitz M, Schackert HK, Engel C, Büttner R, Wijnen J, Doerks T, Bork P, Moebus S, Herms S, Fischer S, Hoffmann P, Aretz S, Steinke-Lange V.

Int J Cancer. 2018;143(11):2800-2813. doi:[10.1002/ijc.31725](https://doi.org/10.1002/ijc.31725)

No Difference in Colorectal Cancer Incidence or Stage at Detection by Colonoscopy Among 3 Countries With Different Lynch Syndrome Surveillance Policies.

Engel C, Vasen HF, Seppälä T, Aretz S, Bigirwamungu-Bargeman M, de Boer SY, Bucksch K, Büttner R, Holinski-Feder E, Holzapfel S, Hüneburg R, Jacobs MAJM, Järvinen H, Kloor M, von Knebel Doeberitz M, Koornstra JJ, van Kouwen M, Langers AM, van de Meeberg PC, Morak M, Möslein G, Nagengast FM, Pylvänäinen K, Rahner N, Renkonen-Sinisalo L, Sanduleanu S, Schackert HK, Schmiegel W, Schulmann K, Steinke-Lange V, Strassburg CP, Vecht J, Verhulst M-L, de Vos Tot Nederveen Cappel W, Zachariae S, Mecklin J-P, Loeffler M.

Gastroenterology. 2018;155(5):1400-1409.e2. doi:[10.1053/j.gastro.2018.07.030](https://doi.org/10.1053/j.gastro.2018.07.030)

Interdisciplinary Diagnosis, Therapy and Follow-up of Patients with Endometrial Cancer. Guideline (S3-Level, AWMF Registry Number 032/034-OL, April 2018) - Part 2 with Recommendations on the Therapy and Follow-up of Endometrial Cancer, Palliative Care, Psycho-oncological/Psychosocial Care/Rehabilitation/Patient Information and Healthcare Facilities.

Emons G, Steiner E, Vordermark D, Uleer C, Bock N, Paradies K, Ortman O, Aretz S, Mallmann P, Kurzeder C, Hagen V, van Oorschot B, Höcht S, Feyer P, Egerer G, Friedrich M, Cremer W, Prott F-J, Horn L-C, Prömpeler H, Langrehr J, Leinung S, Beckmann MW, Kimmig R, Letsch A, Reinhardt M, Alt-Epping B, Kiesel L, Menke J, Gebhardt M, Steinke-Lange V, Rahner N, Lichtenegger W, Zeimet A, Hanf V, Weis J, Mueller M, Henschler U, Schmutzler RK, Meindl A, Hilpert F, Panke JE, Strnad V, Niehues C, Dauelsberg T, Niehoff P, Mayr D, Grab D, Kreißl M, Witteler R, Schorsch A, Mustea A, Petru E, Hübner J, Rose AD, Wight E, Tholen R, Bauerschmitz GJ, Fleisch M, Juhasz-Boess I, Lax S, Runnebaum I, Tempfer C, Nothacker MJ, Blödt S, Follmann M, Langer T, Raatz H, Wesselmann S, Erdogan S. *Geburtshilfe Frauenheilkd.* 2018;78(11):1089-1109. doi:[10.1055/a-0715-2964](https://doi.org/10.1055/a-0715-2964)

Cancer Risks for PMS2-Associated Lynch Syndrome.

Ten Broeke SW, van der Klift HM, Tops CMJ, Aretz S, Bernstein I, Buchanan DD, de la Chapelle A, Capella G, Clendenning M, Engel C, Gallinger S, Gomez Garcia E, Figueiredo JC, Haile R, Hampel HL, Hopper JL, Hoogerbrugge N, von Knebel Doeberitz M, Le Marchand L, Letteboer TGW, Jenkins MA, Lindblom A, Lindor NM, Mensenkamp AR, Møller P, Newcomb PA, van Os TAM, Pearlman R, Pineda M, Rahner N, Redeker EJW, Olderoode-Berends MJW, Rosty C, Schackert HK, Scott R, Senter L, Spruijt L, Steinke-Lange V, Suerink M, Thibodeau S, Vos YJ, Wagner A, Winship I,

Hes FJ, Vasen HFA, Wijnen JT, Nielsen M, Win AK.
J Clin Oncol. 2018;36(29):2961-2968. doi:[10.1200/JCO.2018.78.4777](https://doi.org/10.1200/JCO.2018.78.4777)

Interdisciplinary Diagnosis, Therapy and Follow-up of Patients with Endometrial Cancer. Guideline (S3-Level, AWMF Registry Nummer 032/034-OL, April 2018) - Part 1 with Recommendations on the Epidemiology, Screening, Diagnosis and Hereditary Factors of Endometrial Cancer.

Emons G, Steiner E, Vordermark D, Uleer C, Bock N, Paradies K, Ortman O, Aretz S, Mallmann P, Kurzeder C, Hagen V, van Oorschot B, Höcht S, Feyer P, Egerer G, Friedrich M, Cremer W, Prött F-J, Horn L-C, Prömpeler H, Langrehr J, Leinung S, Beckmann MW, Kimmig R, Letsch A, Reinhardt M, Alt-Epping B, Kiesel L, Menke J, Gebhardt M, Steinke-Lange V, Rahner N, Lichtenegger W, Zeimet A, Hanf V, Weis J, Mueller M, Henschler U, Schmutzler RK, Meindl A, Hilpert F, Panke JE, Strnad V, Niehues C, Dauelsberg T, Niehoff P, Mayr D, Grab D, Kreißl M, Witteler R, Schorsch A, Mustea A, Petru E, Hübner J, Rose AD, Wight E, Tholen R, Bauerschmitz GJ, Fleisch M, Juhasz-Boess I, Sigurd L, Runnebaum I, Tempfer C, Nothacker MJ, Blödt S, Follmann M, Langer T, Raatz H, Wesselmann S, Erdogan S.
Geburtshilfe Frauenheilkd. 2018;78(10):949-971. doi:[10.1055/a-0713-1218](https://doi.org/10.1055/a-0713-1218)

2015

Frequency and phenotypic spectrum of germline mutations in *POLE* and seven other polymerase genes in 266 patients with colorectal adenomas and carcinomas: *POLE* Mutations in Polyposis.

Spier I, Holzapfel S, Altmüller J, Zhao B, Horpaopan S, Vogt S, Chen S, Morak M, Raeder S, Kayser K, Stienen D, Adam R, Nürnberg P, Plotz G, Holinski-Feder E, Lifton RP, Thiele H, Hoffmann P, Steinke V, Aretz S.
Int J Cancer. 2015;137(2):320-331. doi:[10.1002/ijc.29396](https://doi.org/10.1002/ijc.29396)

Genome-wide CNV analysis in 221 unrelated patients and targeted high-throughput sequencing reveal novel causative candidate genes for colorectal adenomatous polyposis: CNV analysis polyposis.

Horpaopan S, Spier I, Zink AM, Altmüller J, Holzapfel S, Laner A, Vogt S, Uhlhaas S, Heilmann S, Stienen D, Pastermack SM, Keppler K, Adam R, Kayser K, Moebus S, Draaken M, Degenhardt F, Engels H, Hofmann A, Nöthen MM, Steinke V, Perez-Bouza A, Herms S, Holinski-Feder E, Fröhlich H, Thiele H, Hoffmann P, Aretz S.
Int J Cancer. 2015;136(6):E578-E589. doi:[10.1002/ijc.29215](https://doi.org/10.1002/ijc.29215)

Functional testing strategy for coding genetic variants of unclear significance in *MLH1* in Lynch syndrome diagnosis.

Hinrichsen I, Schafer D, Langer D, Koger N, Wittmann M, Aretz S, Steinke V, Holzapfel S, Trojan J, König R, Zeuzem S, Brieger A, Plotz G. *Carcinogenesis.* 2015;36(2):202-211. doi:[10.1093/carcin/bgu239](https://doi.org/10.1093/carcin/bgu239)

2014

Germline variants in the *SEMA4A* gene predispose to familial colorectal cancer type X.

Schulz E, Klampfl P, Holzapfel S, Janecke AR, Ulz P, Renner W, Kashofer K, Nojima S, Leitner A, Zebisch A, Wölfler A, Hofer S, Gerger A, Lax S, Beham-Schmid C, Steinke V, Heitzer E, Geigl JB, Windpassinger C, Hoeffler G, Speicher MR, Richard Boland C, Kumanogoh A, Sill H. *Nat Commun.* 2014;5(1):5191. doi:[10.1038/ncomms6191](https://doi.org/10.1038/ncomms6191)

Evaluating the performance of clinical criteria for predicting mismatch repair gene mutations in Lynch syndrome: A comprehensive analysis of 3,671 families: Performance of clinical criteria for Lynch syndrome.

Steinke V, Holzapfel S, Loeffler M, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Pox C, Royer-Pokora B, von Knebel-Doerberitz M, Büttner R, Propping P, Engel C, on behalf of the German HNPCC Consortium.
Int J Cancer. 2014;135(1):69-77. doi:[10.1002/ijc.28650](https://doi.org/10.1002/ijc.28650)

Genome-wide analysis associates familial colorectal cancer with increases in copy number variations and a rare structural variation at 12p12.3.

Yang R, Chen B, Pfütze K, Buch S, Steinke V, Holinski-Feder E, Stocker S, von Schonfels W, Becker T, Schackert HK, Royer-Pokora B, Kloor M, Schmiegel WH, Büttner R, Engel C, Lascorz Puertolas J, Forsti A, Kunkel N, Bugert P, Schreiber S, Krawczak M, Schafmayer C, Propping P, Hampe J, Hemminki K, Burwinkel B.
Carcinogenesis. 2014;35(2):315-323. doi:[10.1093/carcin/bgt344](https://doi.org/10.1093/carcin/bgt344)

Reduced migration of *MLH1* deficient colon cancer cells depends on *SPTAN1*.

Hinrichsen I, Ernst B, Nuber F, Passmann S, Schäfer D, Steinke V, Friedrichs N, Plotz G, Zeuzem S, Brieger A.
Mol Cancer. 2014;13(1):11. doi:[10.1186/1476-4598-13-11](https://doi.org/10.1186/1476-4598-13-11)

2013

Role of the Oxidative DNA Damage Repair Gene OGG1 in Colorectal Tumorigenesis. Smith CG, West H, Harris R, Idziaszczyk S, Maughan TS, Kaplan R, Richman S, Quirke P, Seymour M, Moskvina V, **Steinke V**, Propping P, Hes FJ, Wijnen J, Cheadle JP. *JNCI: Journal of the National Cancer Institute*. 2013;105(16):1249-1253.
doi:[10.1093/jnci/djt183](https://doi.org/10.1093/jnci/djt183)

Hereditary Nonpolyposis Colorectal Cancer (HNPCC)/Lynch Syndrome. **Steinke V**, Engel C, Büttner R, Schackert HK, Schmiegel WH, Propping P. *Deutsches Ärzteblatt Online*. Published online January 18, 2013.
doi:[10.3238/arztebl.2013.0032](https://doi.org/10.3238/arztebl.2013.0032)

2012

Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2, EPCAM) - update 2012. Rahner N, **Steinke V**, Schlegelberger B, Eisinger F, Hutter P, Olschwang S. *Eur J Hum Genet*. 2013;21(1):118-118.
doi:[10.1038/ejhg.2012.164](https://doi.org/10.1038/ejhg.2012.164)

Risks of Less Common Cancers in Proven Mutation Carriers With Lynch Syndrome. Engel C, Loeffler M, **Steinke V**, Rahner N, Holinski-Feder E, Dietmaier W, Schackert HK, Goergens H, von Knebel Doeberitz M, Goecke TO, Schmiegel W, Buettner R, Moeslein G, Letteboer TGW, García EG, Hes FJ, Hoogerbrugge N, Menko FH, van Os TAM, Sijmons RH, Wagner A, Kluijdt I, Propping P, Vasen HFA. *JCO*. 2012;30(35):4409-4415.
doi:[10.1200/JCO.2012.43.2278](https://doi.org/10.1200/JCO.2012.43.2278)

Mutation and association analyses of the candidate genes ESR1, ESR2, MAX, PCNA, and KAT2A in patients with unexplained MSH2-deficient tumors. The Dutch Cancer Genetics Group, Rahner N, Brockschmidt FF, **Steinke V**, Kahl P, Becker T, Vasen HFA, Wijnen JT, Tops CJM, Holinski-Feder E, Ligtenberg MJL, Spruijt L, Görgens H, Stemmler S, Kloor M, Dietmaier W, Schumacher J, Nöthen MM, Propping P. *Familial Cancer*. 2012;11(1):19-26.
doi:[10.1007/s10689-011-9489-z](https://doi.org/10.1007/s10689-011-9489-z)

2011

FHL2 expression in peritumoural fibroblasts correlates with lymphatic metastasis in sporadic but not in HNPCC-associated colon cancer. Gullotti L, Czerwitzki J, Kirfel J, Propping P, Rahner N, **Steinke V**, Kahl P, Engel C, Schüle R, Buettner R, Friedrichs N. *Lab Invest*. 2011;91(12):1695-1705. doi:[10.1038/labinvest.2011.109](https://doi.org/10.1038/labinvest.2011.109)

Association Between TAS2R38 Gene Polymorphisms and Colorectal Cancer Risk: A Case-Control Study in Two Independent Populations of Caucasian Origin. Carrai M, **Steinke V**, Vodicka P, Pardini B, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Stemmler S, Betz B, Kloor M, Engel C, Büttner R, Naccarati A, Vodickova L, Novotny J, Stein A, Hemminki K, Propping P, Försti A, Canzian F, Barale R, Campa D, Song Y, ed. *PLoS ONE*. 2011;6(6):e20464. doi:[10.1371/journal.pone.0020464](https://doi.org/10.1371/journal.pone.0020464)

Missense variants in hMLH1 identified in patients from the German HNPCC consortium and functional studies. Peter Propping and the German HNPCC consortium, Hardt K, Heick SB, Betz B, Goecke T, Yazdanparast H, Küppers R, Servan K, **Steinke V**, Rahner N, Morak M, Holinski-Feder E, Engel C, Möslein G, Schackert H-K, von Knebel Doeberitz M, Pox C, Hegemann JH, Royer-Pokora B. *Familial Cancer*. 2011;10(2):273-284.
doi:[10.1007/s10689-011-9431-4](https://doi.org/10.1007/s10689-011-9431-4)

Recurrence and variability of germline EPCAM deletions in Lynch syndrome. Kuiper RP, Vissers LELM, Venkatchalam R, Bodmer D, Hoenselaar E, Goossens M, Haufe A, Kamping E, Niessen RC, Hogervorst FBL, Gille JJP, Redeker B, Tops CMJ, van Gijn ME, van den Ouweland AMW, Rahner N, **Steinke V**, Kahl P, Holinski-Feder E, Morak M, Kloor M, Stemmler S, Betz B, Hutter P, Bunyan DJ, Syngal S, Culver JO, Graham T, Chan TL, Nagtegaal ID, van Krieken JHJM, Schackert HK, Hoogerbrugge N, van Kessel AG, Ligtenberg MJL. *Hum Mutat*. 2011;32(4):407-414.
doi:[10.1002/humu.21446](https://doi.org/10.1002/humu.21446)

Risk of colorectal and endometrial cancers in EPCAM deletion-positive Lynch syndrome: a cohort study.

Kempers MJ, Kuiper RP, Ockeloen CW, Chappuis PO, Hutter P, Rahner N, Schackert HK, **Steinke V**, Holinski-Feder E, Morak M, Kloor M, Büttner R, Verziel ET, van Krieken JH, Nagtegaal ID, Goossens M, van der Post RS, Niessen RC, Sijmons RH, Kluijt I, Hogervorst FB, Leter EM, Gille JJ, Aalfs CM, Redeker EJ, Hes FJ, Tops CM, van Nesselrooij BP, van Gijn ME, García EBG, Eccles DM, Bunyan DJ, Syngal S, Stoffel EM, Culver JO, Palomares MR, Graham T, Velsher L, Papp J, Oláh E, Chan TL, Leung SY, van Kessel AG, Kiemeny LA, Hoogerbrugge N, Ligtenberg MJ. *The Lancet Oncology*. 2011;12(1):49-55. doi:[10.1016/S1470-2045\(10\)70265-5](https://doi.org/10.1016/S1470-2045(10)70265-5)

2010

Polymorphisms of genes coding for ghrelin and its receptor in relation to colorectal cancer risk: a two-step gene-wide case-control study.

Campa D, Pardini B, Naccarati A, Vodickova L, Novotny J, **Steinke V**, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Kötting J, Betz B, Kloor M, Engel C, Büttner R, Propping P, Försti A, Hemminki K, Barale R, Vodicka P, Canzian F. *BMC Gastroenterol*. 2010;10(1):112. doi:[10.1186/1471-230X-10-112](https://doi.org/10.1186/1471-230X-10-112)

Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2). Rahner N, **Steinke V**, Schlegelberger B, Olschwang S, Eisinger F, Hutter P. *Eur J Hum Genet*. 2010;18(9):1071-1071. doi:[10.1038/ejhg.2009.232](https://doi.org/10.1038/ejhg.2009.232)

Genome-wide association study for colorectal cancer identifies risk polymorphisms in German familial cases and implicates MAPK signalling pathways in disease susceptibility.

Lascorz J, Försti A, Chen B, Buch S, **Steinke V**, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Schulmann K, Goecke T, Kloor M, Engel C, Büttner R, Kunkel N, Weires M, Hoffmeister M, Pardini B, Naccarati A, Vodickova L, Novotny J, Schreiber S, Krawczak M, Bröring CD, Völzke H, Schafmayer C, Vodicka P, Chang-Claude J, Brenner H, Burwinkel B, Propping P, Hampe J, Hemminki K. *Carcinogenesis*. 2010;31(9):1612-1619. doi:[10.1093/carcin/bgq146](https://doi.org/10.1093/carcin/bgq146)

Efficacy of Annual Colonoscopic Surveillance in Individuals With Hereditary Nonpolyposis Colorectal Cancer.

Engel C, Rahner N, Schulmann K, Holinski-Feder E, Goecke TO, Schackert HK, Kloor M, **Steinke V**, Vogelsang H, Möslin G, Görgens H, Dechant S, von Knebel Doeberitz M, Rüschoff J, Friedrichs N, Büttner R, Loeffler M, Propping P, Schmiegel W. *Clinical Gastroenterology and Hepatology*. 2010;8(2):174-182. doi:[10.1016/j.cgh.2009.10.003](https://doi.org/10.1016/j.cgh.2009.10.003)

2009

Expanded Extracolonic Tumor Spectrum in MUTYH-Associated Polyposis.

Vogt S, Jones N, Christian D, Engel C, Nielsen M, Kaufmann A, **Steinke V**, Vasen HF, Propping P, Sampson JR, Hes FJ, Aretz S. *Gastroenterology*. 2009;137(6):1976-1985.e10. doi:[10.1053/j.gastro.2009.08.052](https://doi.org/10.1053/j.gastro.2009.08.052)

Deciphering the genetics of hereditary non-syndromic colorectal cancer.

The CORGI Consortium, Papaemmanuil E, Carvajal-Carmona L, Sellick GS, Kemp Z, Webb E, Spain S, Sullivan K, Barclay E, Lubbe S, Jaeger E, Vijaya-krishnan J, Broderick P, Gorman M, Martin L, Lucassen A, Bishop DT, Evans DG, Maher ER, **Steinke V**, Rahner N, Schackert HK, Goecke TO, Holinski-Feder E, Propping P, Van Wezel T, Wijnen J, Cazier J-B, Thomas H, Houlston RS, Tomlinson I. *Eur J Hum Genet*. 2008;16(12):1477-1486. doi:[10.1038/ejhg.2008.129](https://doi.org/10.1038/ejhg.2008.129)

Hereditary Cancer Syndromes. Rahner N, **Steinke V**. *Deutsches Ärzteblatt Online*. Published online October 10, 2008. doi:[10.3238/arztebl.2008.0706](https://doi.org/10.3238/arztebl.2008.0706)

Compound heterozygosity for twoMSH6 mutations in a patient with early onset colorectal cancer, vitiligo and systemic lupus erythematosus.

Rahner N, Höefler G, Högenauer C, Lackner C, **Steinke V**, Sengteller M, Friedl W, Aretz S, Propping P, Mangold E, Walldorf C. *Am J Med Genet*. 2008;146A(10):1314-1319. doi:[10.1002/ajmg.a.32210](https://doi.org/10.1002/ajmg.a.32210)

No association between MUTYH and MSH6 germline mutations in 64 HNPCC patients.

for The German HNPCC Consortium, **Steinke V**, Rahner N, Morak M, Keller G, Schackert HK, Görgens H, Schmiegel W, Royer-Pokora B, Dietmaier W, Kloor M, Engel C, Propping P, Aretz S. *Eur J Hum Genet*. 2008;16(5):587-592. doi:[10.1038/ejhg.2008.26](https://doi.org/10.1038/ejhg.2008.26)

Nine novel pathogenic germline mutations in *MLH1*, *MSH2*, *MSH6* and *PMS2* in families with Lynch syndrome. Rahner N, Friedrichs N, Wehner M, **Steinke V**, Aretz S, Friedl W, Buettner R, Mangold E, Propping P, Wall-dorf C. *Acta Oncologica*. 2007;46(6):763-769. doi:[10.1080/02841860701230217](https://doi.org/10.1080/02841860701230217)