

2026

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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.

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Reply: An epigenetic basis for genetic anticipation in facioscapulohumeral muscular dystrophy type 1.

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Reply: An epigenetic basis for genetic anticipation in facioscapulohumeral muscular dystrophy type 1.

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

Yasmin Soares de Lima, Coral Arnau-Collell, Jenifer Muñoz, Cristina Herrera-Pariente, Leticia Moreira, Teresa Ocaña, Marcos Díaz-Gay, Sebastià Franch-Expósito, Miriam Cuatrecasas, Sabela Carballal, Anael Lopez-Novo, Lorena Moreno, Guerau Fernández, Aranzazu Díaz de Bustamante, Sophia Peters, Anna K Sommer, Isabel Spier, Iris B A W Te Paske, Yasmijn J van Herwaarden, Antoni Castells, Luis Bujanda, Gabriel Capellà, Verena Steinke-Lange, Khalid Mahmood, JiHoon Eric Joo, Julie Arnold, Susan Parry, Finlay A Macrae, Ingrid M Winship, Christophe

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Diagnostic yield and clinical relevance of expanded germline genetic testing for nearly 7000 suspected HBOC patients.

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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.

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Twist exome capture allows for lower average sequence coverage in clinical exome sequencing

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Parallel in-depth analysis of repeat expansions in ataxia patients by long-read sequencing

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Methylation of the 4q35 D4Z4 repeat defines disease status in facioscapulohumeral muscular dystrophy

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Clinical, genetic, epidemiologic, evolutionary, and functional delineation of TSPEAR-related autosomal recessive ectodermal dysplasia 14

Adam Jackson, Sheng-Jia Lin, Elizabeth A Jones, Kate E Chandler, David Orr, Celia Moss, Zahra Haider, Gavin Ryan, Simon Holden, Mike Harrison, Nigel Burrows, Wendy D Jones, Mary Loveless, Cassidy Petree, Helen Stewart, Karen Low, Deirdre Donnelly, Simon Lovell, Konstantina Drosou; Genomics England Research Consortium; Solve-RD consortium; Gaurav K Varshney, Siddharth Banka.

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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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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.

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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, Líliliana 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, Nicoline Hoogerbrugge, Carla Oliveira

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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

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Parallel in-depth analysis of repeat expansions in ataxia patients by long-read sequencing

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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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Methylation of the 4q35 D4Z4 repeat defines disease status in facioscapulohumeral muscular dystrophy

Hannes Erdmann, Florentine Scharf, Stefanie Gehling, Anna Benet-Pagès, Sibylle Jakubiczka, Kerstin Becker, Maria Seipelt, Felix Kleefeld, Karl Christian Knop, Eva Christina Prott, Miriam Hiebeler, Federica Montagnese, Dieter Gläser, Matthias Vorgerd, Tim Hagenacker, Maggie C Walter, Peter Reilich, Teresa Neuhan, Martin Zenker, Elke Holinski-Feder, Benedikt Schoser, Angela Abicht

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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

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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 Chauris, Elke Holinski-Feder, Gabriel Capellá

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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

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Metabolic targeting of cancer by a ubiquinone uncompetitive inhibitor of mitochondrial complex I

Shashi Jain, Cheng Hu, Jerome Kluza, Wei Ke, Guiyou Tian, Madalina Giurgiu, Andreas Bleilevens, Alexandre Rosa Campos, Adriana Charbono, Elmar Stickeler, Jochen Maurer, Elke Holinski-Feder, Arkadii Vaisburg, Matthias Bureik, Guangcheng Luo, Philippe Marchetti, Yabin Cheng, Dieter A Wolf

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

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Solving patients with rare diseases through programmatic reanalysis of genome-phenome data

Leslie Matalonga, Carles Hernández-Ferrer, Davide Piscia, Solve-RD SNV-indel working group; Rebecca Schüle, Matthis Synofzik, Ana Töpf, Lisenka E L M Vissers, Richarda de Voer, Solve-RD DITF-GENTURIS; Solve-RD DITF-ITHACA; Solve-RD DITF-euroNMD; Solve-RD DITF-RND; Raul Tonda, Steven Laurie, Marcos Fernandez-Callejo, Daniel Picó, Carles Garcia-Linares, Anastasios Papakonstantinou, Alberto Corvó, Ricky Joshi, Hector Diez, Ivo Gut, Alexander Hoischen, Holm Graessner, Sergi Beltran, Solve-RD Consortia
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<https://doi.org/10.1016/j.ejmg.2022.104475>

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>

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, Maike de Wit, Christopher Haberl, Elke Holinski-Feder, Julia M A Pickl
<https://doi.org/10.3390/cancers14030851>

Initial Clinical Experience with NIPT for Rare Autosomal Aneuploidies and Large Copy Number Variations

Thomas Harasim, Teresa Neuhann, Anne Behnecke, Miriam Stampfer, Elke Holinski-Feder, Angela Abicht
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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 Schröck, Elke Holinski-Feder
<https://doi.org/10.1055/a-1541-7912>

2021

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

Karolin Bucksch, Silke Zachariae, Aysel Ahadova, Stefan Aretz, Reinhard Büttner, Heike Görgens, Elke Holinski-Feder, Robert Hüneburg, Matthias Kloor, Magnus von Knebel Doeberitz, Svetlana Ladigan-Badura, Gabriela Moeslein, Monika Morak, Jacob Nattermann, Huu Phuc Nguyen, Claudia Perne, Silke Redler, Ariane Schmetz, Verena Steinke-Lange, Harald Surowy, Deepak B Vangala, Jürgen Weitz, Markus Loeffler, Christoph Engel, German Consortium for Familial Intestinal Cancer
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<https://doi.org/10.1136/jmedgenet-2021-108147>

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HPO – driven virtual gene panel: a new efficient approach in molecular autopsy of sudden unexplained death

Ulrike Schön, Anna Holzer, Andreas Laner, Stephanie Kleinle, Florentine Scharf, Anna Benet-Pagès, Oliver Peschel, Elke Holinski-Feder, Isabel Diebold
<https://doi.org/10.1186/s12920-021-00946-7>

Early detection of duodenal cancer by upper gastrointestinal – endoscopy in Lynch syndrome

Deepak B Vangala, Svetlana Ladigan-Badura, Christoph Engel, Robert Hüneburg, Claudia Perne, Karolin Bucksch, Jacob Nattermann, Verena Steinke-Lange, Nils Rahner, Jürgen Weitz, Matthias Kloor, Judith Tomann, Ali Canbay, Huu-Phuc Nguyen, Christian Strassburg, Gabriele Möslein, Monika Morak, Elke Holinski-Feder, Reinhard Büttner, Stefan Aretz, Markus Löffler, Wolff Schmiegel, Christian Pox, Karsten Schulmann, German Consortium for Familial Intestinal Cancer
<https://doi.org/10.1002/ijc.33753>

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

Toni T Seppälä, Mev Dominguez-Valentin, Emma J Crosbie, Christoph Engel, Stefan Aretz, Finlay Macrae, Ingrid Winship, Gabriel Capella, Huw Thomas, Eivind Hovig, Maartje Nielsen, Rolf H Sijmons, Lucio Bertario, Bernardo Bonanni, Maria G Tibiletti, Giulia M Cavestro, Miriam Mints, Nathan Gluck, Lior Katz, Karl Heinimann, Carlos A Vaccaro, Kate Green, Fiona Laloo, James Hill, Wolff Schmiegel, Deepak Vangala, Claudia Perne, Hans-Georg Strauß, Johanna Tecklenburg, Elke Holinski-Feder, Verena Steinke-Lange, Jukka-Pekka Mecklin, John-Paul Plazzer, Marta Pineda, Matilde Navarro, Joan B Vida, Revital Kariv, Guy Rosner, Tamara A Piñero, Walter Pavicic, Pablo Kalfayan, Sanne W Ten Broeke, Mark A Jenkins, Lone Sunde, Inge Bernstein, John Burn, Marc Greenblatt, Wouter H de Vos Tot Nederveen Cappel, Adriana Della Valle, Francisco Lopez-Koestner, Karin Alvarez, Reinhard Büttner, Heike Görgens, Monika Morak, Stefanie Holzapfel, Robert Hüneburg, Magnus von Knebel Doeberitz, Markus Loeffler, Silke Redler, Jürgen Weitz, Kirsi Pylvänäinen, Laura Renkonen-Sinisalo, Anna Lepistö, John L Hopper, Aung K Win, Noralane M Lindor, Steven Gallinger, Loïc Le Marchand, Polly A Newcomb, Jane C Figueiredo, Stephen N Thibodeau, Christina Therkildsen, Karin A W Wadt, Marian J E Mourits, Zohreh Ketabi, Oliver G Denton, Einar A Rødland, Hans Vasen, Florencia Neffa, Patricia Esperon, Douglas Tjandra, Gabriela Möslein, Erik Rökkones, Julian R Sampson, D G Evans, Pål Møller
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<https://doi.org/10.1016/j.ejmg.2021.104360>

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<https://doi.org/10.1002/ijc.33790>

Correction: Solving unsolved rare neurological diseases –a Solve-RD viewpoint

Rebecca Schüle, Dagmar Timmann, Corrie E Erasmus, Jennifer Reichbauer, Melanie Wayand, Solve-RD-DITF-RND; Bart van de Warrenburg, Ludger Schöls, Carlo Wilke, Andrea Bevot, Stephan Zuchner, Sergi Beltran, Steven Laurie, Leslie Matalonga, Holm Graessner, Matthis Synofzik, Solve-RD Consortium
<https://doi.org/10.1038/s41431-021-00935-5>

Correction to: Solving patients with rare diseases through programmatic reanalysis of genome-phenome data

Leslie Matalonga, Carles Hernández-Ferrer, Davide Piscia, Solve-RD SNV-indel working group; Rebecca Schüle, Matthis Synofzik, Ana Töpf, Lisenka E L M Vissers, Richarda de Voer, Solve-RD DITF-GENTURIS; Solve-RD DITF-ITHACA; Solve-RD DITF-euroNMD; Solve-RD DITF-RND; Raul Tonda, Steven Laurie, Marcos Fernandez-Callejo, Daniel Picó, Carles Garcia-Linares, Anastasios Papakonstantinou, Alberto Corvó, Ricky Joshi, Hector Diez, Ivo Gut, Alexander Hoischen, Holm Graessner, Sergi Beltran, Solve-RD Consortium
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