

## 2026

### **Hereditary cancer: Germline testing practices across ERN GENTURIS member countries.**

Kiljańczyk M, Daneberga Z, Tooming M, Urbańczyk K, Pöyhönen M, Kahre T, Foretova L, Tham E, Milagre T, Melegh B, Haanpää MK, Blatnik A, Wimmer K, de Putter R, Wadt K, Houdayer C, Holinski-Feder E, Kattamis A, Klink B, Høberg-Vetti H, Blanco Guillermo I, Hoogerbrugge N, Lubiński J.

<https://pubmed.ncbi.nlm.nih.gov/42265275/>

### **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 (Holinski-Feder 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, Möslein 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/>

### **Validation structures for sequence variants of uncertain significance in hereditary cancer.**

Lucas MC, Keßler T, Benet-Pagès A, Holinski-Feder E, Laner A, Klink B.

<https://pubmed.ncbi.nlm.nih.gov/41807735/>

### **Repeat-associated ataxias in a German patient cohort analysed by targeted parallel long-read sequencing.**

Erdmann H, Schaub A, Lucas MC, Scholz V, Benet-Pages A, Becker K, Dineiger C, Mayer V, van Buren I, Breithausen E, Akbari K, Cordts I, Sauer M, Schneider C, Krakowsky R, Schnabel F, Dunker K, Fabritius L, Gerb J, Grabova D, Möhwald K, Näher M, Steinmetz K, Thiessen F, Jäck A, Schneider-Gold C, Zittel S, Petersen C, Schreyer I, Mämecke L, Wilfling S, Wunderlich G, Brenner D, Hellenbroich Y, Muhle K, Huchtemann T, Claus I, Klopstock T, Strupp M, Levin J, Höglinger G, Huppert D, Becker-Bense S, Filippoulos F, Kilpert F, Leitão E, Kaya S, Depienne C, Schöberl F, Neuhann T, Holinski-Feder E, Zwergal A, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/40898875/>

### **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/>

### **Next-Generation Sequencing in Congenital Eye Malformations: Identification of Genetic Causes and Comparison of Different Panel-Based Diagnostic Strategies.**

Neuhann L, Laner A, Holinski-Feder E, Neuhann T.

<https://pubmed.ncbi.nlm.nih.gov/41155148/>

### **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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**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/>

**Umfassende Charakterisierung des D4Z4-Repeatarrays mittels Long-Read-Sequenzierung für eine präzise Diagnostik der Fazioskapulohumeralen Muskeldystrophie.**

Scharf F, Erdmann H, Lucas MC, Gehling S, Benet-Pagès A, Schäfer J, Hallermayr A, Schönrock V, Köhler U, Neuhann T, Holinski-Feder E, Walter M, Schoser B, Abicht A.

Nervenheilkunde 2025; 44(03): 168-169. DOI: [10.1055/s-0044-1801528](https://doi.org/10.1055/s-0044-1801528)

**Die Ränder des diagnostischen Spektrums der FSHD – Komplexe genetische Befunde in der Diagnostik der Fazioskapulohumeralen Muskeldystrophie (FSHD) und ihre Implikation für das molekulargenetische Modell der Erkrankung.**

Erdmann H, Gehling S, Scharf F, Lucas MC, Kleefeld F, Becker K, Schönrock V, Saak A, Schäfer J, Neuhoff S, Hagenacker T, Pormann J, Rausch HW, Schirmer L, Berking AC, Neuhann T, Holinski-Feder E, Walter M, Schoser B, Abicht A.

Nervenheilkunde 2025; 44(03): 143. DOI: [10.1055/s-0044-1801447](https://doi.org/10.1055/s-0044-1801447)

**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

**Parallel in-depth analysis of repeat expansions: an updated Clin-CATS workflow for nanopore R10 flow cells.**

Scholz V, Schönrock V, Erdmann H, Prokosch V, Schoedel M, Almus M, Sauer M, Mayer V, Breithausen E, van Buren I, Dineiger C, Heintz C, Hallermayr A, Neuhann T, Holinski-Feder E, Abicht A, Benet-Pagès A, Lucas MC.

bioRxiv 2024.11.05.622099; doi: <https://doi.org/10.1101/2024.11.05.622099>

**Updated Structure of CNBP Repeat Expansions in Patients With Myotonic Dystrophy Type 2 and Its Implication for Standard Diagnostics.**

Wendlandt M, Erdmann H, Rost S, Lucas MC, Becker K, Kleinle S, Timmer M, Bier A, Wunderlich G, Wenninger S, Walter MC, Neuhann T, Schoser B, Holinski-Feder E, Abicht A.

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**Eccentric hypertrophy impairs outcome after TAVR.**

Thalmann R, Obermeier V, Westphal DS, Diebold I, Trenkwalder T, Pellegrini C, Buglio G, Seoudy H, Hoppmann P, Bradaric C, Schön U, Holinski-Feder E, Lettmann N, Ruge H, Erlebach M, Fuetterer C, Laugwitz KL, Krane M, Frank D, Kupatt C.

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**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 (u. a. Holinski-Feder E, Laner A, Steinke-Lange V et al.), Laurie S.

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**[Lynch syndrome]**

Steinke-Lange V, Holinski-Feder E.

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

**Optical Genome Mapping as a Potential Routine Clinical Diagnostic Method.**

Barseghyan H, Eisenreich D, Lindt E, Wendlandt M, Scharf F, Benet-Pages A, Sendelbach K, Neuhann T, Abicht A, Holinski-Feder E, Koehler U.

<https://pubmed.ncbi.nlm.nih.gov/38540401/>

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

Johansson LF, Laurie S, Spalding D, Gibson S, Ruvolo 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, Neerincx 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 (u. a. Holinski-Feder E, Laner A, Steinke-Lange V et al.).

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## 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/>

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

Erdmann H, Scharf F, Hallermayr A, Barseghyan H, Walter MC, Holinski-Feder E, Schoser B, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/37348868/8>

**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.

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**Dominantly inherited micro-satellite instable cancer - the four Lynch syndromes - an EHTG, PLSD position statement.**

Møller P, Seppälä TT, Ahadova A, Crosbie EJ, Holinski-Feder E, Scott R, Haupt S, Möslin G, Winship I, Broeke SWB, Kohut KE, Ryan N, Bauerfeind P, Thomas LE, Evans DG, Aretz S, Sijmons RH, Half E, Heinemann K, Horisberger K, Monahan K, Engel C, Cavestro GM, Fruscio R, Abu-Freha N, Zohar L, Laghi L, Bertario L, Bonanni B, Tibiletti MG, Lino-Silva LS, Vaccaro C, Valle AD, Rossi BM, da Silva LA, de Oliveira Nascimento IL, Rossi NT, Dębnia T, Mecklin JP, Bernstein I, Lindblom A, Sunde L, Nakken S, Heuveline V, Burn J, Hovig E, Kloor M, Sampson JR, Dominguez-Valentin M; Prospective Lynch Syndrome Database (www.plsd.eu) and The European Hereditary Tumour Group (www.ehtg.org).

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**Closing the Gap - Detection of 5q-Spinal Muscular Atrophy by Short-Read Next-Generation Sequencing and Unexpected Results in a Diagnostic Patient Cohort.**

Kleinle S, Scholz V, Benet-Pagès A, Wohlfrom T, Gehling S, Scharf F, Rost S, Prött EC, Grinzinger S, Hotter A, Haug V, Niemeier S, Wiethoff-Ubrig L, Hagenacker T, Goldhahn K, von Moers A, Walter MC, Reilich P, Eggermann K, Kraft F, Kurth I, Erdmann H, Holinski-Feder E, Neuhann T, Abicht A.

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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, Capellà G, Valle L.

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

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

Erdmann H, Scharf F, Hallermayr A, Barsegehyan H, Walter MC, Holinski-Feder E, Schoser B, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/37348868/>

**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 Rosty, Joaquin Cubiella, Daniel Rodríguez-Alcalde, Elke Holinski-Feder, Richarda de Voer, Daniel D Buchanan, Stefan Aretz, Clara Ruiz-Ponte, Laura Valle, Francesc Balaguer, Laia Bonjoch, Sergi Castellvi-Bel.

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

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

Burcu Yaldiz, Erdi Kucuk, Juliet Hampstead, Tom Hofste, Rolph Pfundt, Jordi Corominas Galbany, Tuula Rinne, Helger G Yntema, Alexander Hoischen, Marcel Nelen, Christian Gilissen; Solve-RD consortium.

<https://pubmed.ncbi.nlm.nih.gov/37138343/>

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

Hannes Erdmann, Florian Schöberl, Mădălina Giurgiu, Rafaela Magalhaes Leal Silva, Veronika Scholz, Florentine Scharf, Martin Wendlandt, Stephanie Kleinle, Marcus Deschauer, Georg Nübling, Wolfgang Heide, Sait Seymen Babacan, Christine Schneider, Teresa Neuhann, Katrin Hahn, Benedikt Schoser, Elke Holinski-Feder, Dieter A Wolf, Angela Abicht.

<https://pubmed.ncbi.nlm.nih.gov/36227727/>

**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 Neuhann, Martin Zenker, Elke Holinski-Feder, Benedikt Schoser, Angela Abicht.

<https://pubmed.ncbi.nlm.nih.gov/36100962/>

**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.

<https://pubmed.ncbi.nlm.nih.gov/37009414/>

**A Solve-RD ClinVar-based reanalysis of 1522 index cases from ERN-ITHACA reveals common pitfalls and misinterpretations in exome sequencing**

Denommé-Pichon AS, Matalonga L, de Boer E, Jackson A, Benetti E, Banka S, Bruel AL, Ciolfi A, Clayton-Smith J, Dallapiccola B, Duffourd Y, Ellwanger K, Fallerini C, Gilissen C, Graessner H, Haack TB, Havlovicova M, Hoischen A, Jean-Marçais N, Kleefstra T, López-Martín E, Macek M, Mencarelli MA, Moutton S, Pfundt R, Pizzi S, Posada M, Radio FC, Renieri A, Rooryck C, Ryba L, Safradou H, Schwarz M, Tartaglia M, Thauvin-Robinet C, Thevenon J, Tran Mau-Them F, Trimouille A, Votypka P, de Vries BBA, Willemssen MH, Zurek B, Verloes A, Philippe C; Solve-RD DITF-ITHACA; Solve-RD SNV-indel Working Group; Solve-RD Consortia; Orphanomix Group; Vitobello A, Vissers LELM, Faivre L.

<https://pubmed.ncbi.nlm.nih.gov/36681873/>

**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/>

**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.**

Garcia-Pelaez J, Barbosa-Matos R, Lobo S, Dias A, Garrido L, Castedo S, Sousa S, Pinheiro H, Sousa L, Monteiro R, Maqueda JJ, Fernandes S, Carneiro F, Pinto N, Lemos C, Pinto C, Teixeira MR, Aretz S, Bajalica-Lagercrantz S, Balmaña J, Blatnik A, Benusiglio PR, Blanluet M, Bours V, Brems H, Brunet J, Calistri D, Capellá G, Carrera S, Colas C, Dahan K, de Putter R, Desseignés C, Domínguez-Garrido E, Egas C, Evans DG, Feret D, Fewings E, Fitzgerald RC, Coulet F, Garcia-Barcina M, Genuardi M, Golmard L, Hackmann K, Hanson H, Holinski-Feder E, Hüneburg R, Krajc M, Lagerstedt-Robinson K, Lázaro C, Ligtenberg MJL, Martínez-Bouzas C, Merino S, Michils G, Novaković S, Patiño-García A, Ranzani GN, Schröck E, Silva I, Silveira C, Soto JL, Spier I, Steinke-Lange V, Tedaldi G, Tejada MI, Woodward ER, Tischkowitz M, Hoogerbrugge N, Oliveira C.

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

**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, Nicoline 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

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

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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

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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, 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 Chauris, Elke Holinski-Feder, Gabriel Capell a  
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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 patients with rare diseases through programmatic reanalysis of genome-phenome data**

Leslie Matalonga, Carles Hern andez-Ferrer, Davide Piscia, Solve-RD SNV-indel working group; Rebecca Sch ule, Matthis Synofzik, Ana T opf, 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 Pic o, Carles Garcia-Linares, Anastasios Papakonstantinou, Alberto Corv o, Ricky Joshi, Hector Diez, Ivo Gut, Alexander Hoischen, Holm Graessner, Sergi Beltran, Solve-RD Consortia

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**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

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**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

<https://doi.org/10.3390/jcm11020372>

**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

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## 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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### **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>

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<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 Holzappel, 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 Rokkones, Julian R Sampson, D G Evans, Pål Møller

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**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

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**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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**Correction to: 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).

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**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. Lalloo, 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>.

**Beta-2-microglobulin Mutations Are Linked to a Distinct Metastatic Pattern and a Favorable Outcome in Microsatellite-Unstable Stage IV Gastrointestinal Cancers**

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#### **Hereditary non-polyposis tumor risk syndromes**

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#### **A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report.**

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

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