

## 2022

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

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

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

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

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

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

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

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

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

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

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

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### **Somatic mosaics in hereditary tumor predisposition syndromes**

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

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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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## 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

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

### **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>

### **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

<https://doi.org/10.1016/j.chembiol.2021.11.002>

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

Deepak B Vangala, Swetlana 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 Rokkones, Julian R Sampson, D G Evans, Pål Møller

<https://doi.org/10.1016/j.ejca.2021.02.022>

### **Somatic mosaics in hereditary tumor predisposition syndromes**

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

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

### **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, Mösllein 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>

### **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, Büttner R, Görgens H, Holinski-Feder E, Hüneburg 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://pubmed.ncbi.nlm.nih.gov/34469588/>

### **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. Mösllein, 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>.

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

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>

### **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>

### **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. EMourits, Z. Ketabi, O. G. Denton, E. A. Rødland, H. Vasen, F. Neffa, P. Esperon, D. Tjandra, G. Möslein, 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. 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. 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. Möslein, 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. Schmiegel, 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>

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

E. Busch, A. Ahadova, K. Kosmalla, L. Bohaumilitzky, P. L. Pfuderer, A. Ballhausen, J. Witt, J.-N. Wittemann, H. Bläker, E. Holinski-Feder, D. Jäger, M. von Knebel Doeberitz, G. M. Haag, & M. Kloor  
Frontiers in Oncology, 11 (2021) 669774. <https://doi.org/10.3389/fonc.2021.669774>.

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

Ahadova, T. T. Seppälä, C. Engel, R. Gallon, J. Burn, E. Holinski-Feder, V. Steinke-Lange, G. Möslein, M. Nielsen, S. W. Ten Broeke, L. Laghi, M. Dominguez-Valentin, G. Capella, F. Macrae, R. Scott, R. Hüneburg, J. Nattermann, M. Hoffmeister, H. Brenner, H. Bläker, M. von Knebel Doeberitz, J. R. Sampson, H. Vasen, J.-P. Mecklin, P. Møller, & M. Kloor  
International Journal of Cancer, 148 (2021) 800–811. <https://doi.org/10.1002/ijc.33224> .

## Analysis in the Prospective Lynch Syndrome Database identifies sarcoma as part of the Lynch syndrome tumor spectrum.M.

Dominguez-Valentin, J. R. Sampson, P. Møller, T. T. Seppälä, & PLSD Collaborators  
International Journal of Cancer, 148 (2021) 512–513. <https://doi.org/10.1002/ijc.33214>

## 2020

### 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

Christoph Engel, Aysel Ahadova, Toni T. Seppälä, Stefan Aretz, Marloes Bigirwamungu-Bargeman, Hendrik Bläker, Karolin Bucksch, Reinhard Büttner, Wouter T. de Vos tot Nederveen Cappel, Volker Endris, Elke Holinski-Feder, Stefanie Holzapfel, Robert Hüneburg, Maarten A.J.M. Jacobs, Jan J. Koornstra, Alexandra M. Langers, Anna Lepistö, Monika Morak, Gabriela Möslein, Päivi Peltomäki, Kirsi Pylvänäinen, Nils Rahner, Laura Renkonen-Sinisalo, Karsten Schulmann, Verena Steinke-Lange, Albrecht Stenzinger, Christian P. Strassburg, Paul C. van de Meeberg, Mariette van Kouwen, Monique van Leerdam, Deepak B. Vangala, Juda Vecht, Marie-Louise Verhulst, Magnus von Knebel Doeberitz, Jürgen Weitz, Silke Zachariae, Markus Loeffler, Jukka-Pekka Mecklin, Matthias Kloor, Hans F. Vasen *Gastroenterology*. 2020;158(5):1326-1333. doi:10.1053/j.gastro.2019.12.032

### Früherkennung, Risikoreduktion, Überwachung und Therapie bei Patienten mit Lynch-Syndrom

Robert Hüneburg<sup>1, 2</sup>, Stefan Aretz <sup>2, 3</sup>, Reinhard Büttner <sup>4</sup>, Severin Daum<sup>5</sup>, Christoph Engel<sup>6</sup>, Guido Fechner<sup>2, 7</sup>, Jens K. Habermann<sup>8</sup>, Dominik Heling<sup>1, 2</sup>, Katrin Hoffmann<sup>9</sup>, Elke Holinski-Feder<sup>10, 11</sup>, Matthias Kloor <sup>12</sup>, Magnus von Knebel-Döberitz<sup>12</sup>, Markus Loeffler <sup>6</sup>, Gabriela Möslein<sup>13</sup>, Claudia Perne<sup>2, 3</sup>, Silke Redler<sup>14</sup>, Olaf Rieß<sup>15</sup>, Wolff Schmiegel<sup>16</sup>, Thomas Seufferlein <sup>17</sup>, Ulrike Siebers-Renelt<sup>18</sup>, Verena Steinke-Lange <sup>10, 11</sup>, Johanna Tecklenburg<sup>19</sup>, Deepak Vangala<sup>16</sup>, Tim Vilz<sup>2, 20</sup>, Jürgen Weitz<sup>21</sup>, Bertram Wiedenmann<sup>22</sup>, Christian P. Strassburg<sup>1, 2\*</sup>, Jacob Nattermann<sup>1, 2\*</sup> *Gastroenterol* 2019; 57: 1–12. doi.org/10.1055/a-1008-9827

### 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, Therkildsen 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, Möslein G, Mecklin J-P, Nielsen M, Møller P. *Genet Med*. 2020;22(1):15–25.

### 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**, Möslein 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, German HNPCC Consortium, the Dutch Lynch Syndrome Collaborative Group, Finnish Lynch Syndrome Registry.

Gastroenterology. April 2020;158(5):1326–33.

**Analysis of 3297 individuals suggests that the pathogenic germline 5'-UTR variant BRCA1 c.-107A > T is not common in south-east Germany.**

Laner A, Benet-Pages A, Neitzel B, Holinski-Feder E.  
Fam Cancer. 21. März 2020.

**Targeted deep-intronic sequencing in a cohort of unexplained cases of suspected Lynch syndrome.**

Arnold AM, Morak M, Benet-Pagès A, Laner A, Frishman D, Holinski-Feder E. Eur J Hum Genet. 2011 2020;28(5):597-608. doi:10.1038/s41431-019-0536-9

**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

**Cancer incidence and spectrum among children with genetically confirmed Beckwith-Wiedemann spectrum in Germany: a retrospective cohort study.**

Cöktü S, Spix C, Kaiser M, Beygo J, Kleinle S, Bachmann N, Kohlschmidt N, Prawitt D, Beckmann A, Klaes R, Nevinny-Stickel-Hinzpeter C, Döhnert S, Kraus C, Kadgien G, Vater I, Biskup S, Kutsche M, Kohlhase J, Eggermann T, Zenker M, Kratz CP.  
Br J Cancer. 2020;123(4):619-623. doi:10.1038/s41416-020-0911-x

**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, Möslein G, Schackert H-K, Weitz J, Perne C, Aretz S, Hüneburg R, Schmiegel 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

**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, Schmiegel W, Pox C, Schulmann K.  
Int J Cancer. Published online September 15, 2020. doi: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

**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, Möslein 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

**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, Heinemann K, Vaccaro CA, Green K, Laloo F, Hill J, Schmiegel 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 NAJ, Evans DG, Møller P, Crosbie EJ.

**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

**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

**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, Therkildsen 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

**Guidelines for the Li-Fraumeni and heritable TP53-related cancer syndromes.**

T. Frebourg, S. Bajalica Lagercrantz, C. Oliveira, R. Magenheimer, D. G. Evans, & European Reference Network GENTURIS, European journal of human genetics: EJHG, 28 (2020) 1379–1386. <https://doi.org/10.1038/s41431-020-0638-4>.

**Correction: Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database.**

M. Dominguez-Valentin, J. R. Sampson, T. T. Seppälä, S. W. Ten Broeke, J.-P. Plazzer, S. Nakken, C. Engel, S. Aretz, M. A. Jenkins, L. Sunde, I. Bernstein, G. Capella, F. Balaguer, H. Thomas, D. G. Evans, J. Burn, M. Greenblatt, E. Hovig, W. H. de Vos Tot Nederveen Cappel, R. H. Sijmons, L. Bertario, M. G. Tibiletti, G. M. Cavestro, A. Lindblom, A. Della Valle, F. Lopez-Köstner, N. Gluck, L. H. Katz, K. Heinimann, C. A. Vaccaro, R. Büttner, H. Görgens, E. Holinski-Feder, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, N. Rahner, H. K. Schackert, V. Steinke-Lange, W. Schmiegel, D. Vangala, K. Pylvänäinen, L. Renkonen-Sinisalo, 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, K. Wadt, C. Therkildsen, H. Okkels, Z. Ketabi, L. Moreira, A. Sánchez, M. Serra-Burriel, M. Pineda, M. Navarro, I. Blanco, K. Green, F. Laloo, E. J. Crosbie, J. Hill, O. G. Denton, I. M. Frayling, E. A. Rødland, H. Vasen, M. Mints, F. Neffa, P. Esperon, K. Alvarez, R. Kariv, G. Rosner, T. A. Pinero, M. L. Gonzalez, P. Kalfayan, D. Tjandra, I. M. Winship, F. Macrae, G. Möslin, J.-P. Mecklin, M. Nielsen, & P. Møller, Genetics in Medicine: Official Journal of the American College of Medical Genetics, 22 (2020) 1569. <https://doi.org/10.1038/s41436-020-0892-4>

### 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, Schmiegel W, Pox C, Schulmann K. Int J Cancer. Published online September 15, 2020. [https://10.1002/ijc.33294](https://doi.org/10.1002/ijc.33294)

### Contribution of mRNA Splicing to Mismatch Repair Gene Sequence Variant Interpretation.

B. A. Thompson, R. Walters, M. T. Parsons, T. Dumenil, M. Drost, Y. Tiersma, N. M. Lindor, S. V. Tavtigian, N. de Wind, A. B. Spurdle, & InSiGHT Variant Interpretation Committee, Frontiers in Genetics, 11 (2020) 798. <https://doi.org/10.3389/fgene.2020.00798>.

## 2019

### Targeted deep-intronic sequencing in a cohort of unexplained cases of suspected Lynch syndrome.

Arnold AM, **Morak M**, **Benet-Pagès A**, **Laner A**, Frishman D, **Holinski-Feder E**. Eur J Hum Genet. 10. Dezember 2019;

### Results of multigene panel testing in familial cancer cases without genetic cause demonstrated by single gene testing.

Dominguez-Valentin M, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, **Morak M**, **Holinski-Feder E**, Holth A, Capella G, Davidson B, Evans DG, Martins A, Møller P, Hovig E. Sci Rep. 6. Dezember 2019;9(1):18555.

### Das **Birt-Hogg-Dubé-Syndrom**

**Steinke-Lange V**, **Holinski-Feder E**

TZM-News 4/2019, ISSN: 1437-8019

### 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, Moeslein 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.

### Darmkrebs: Vorsorge und Früherkennung neu geregelt

**Gross M**, **Holinski-Feder E**.

MMW Fortschr Med. 2019;161(7):43–8.

### Empfehlungen zur Früherkennung, Risikoreduktion, Überwachung und Therapie bei Patienten mit Lynch-Syndrom

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, Moeslein 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. November 2019;57(11):1309–20.

### 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**.



European Journal of Human Genetics. 22. Juli 2019;1.

### **Genetic Screening and Personalized Prevention in Colorectal Cancer.**

**Steinke-Lange V, Holinski-Feder E.**

VIS. 2019;35(4):226–30.

### **The apparent genetic anticipation in PMS2-associated Lynch syndrome families is explained by birth cohort effect.**

Ten Broeke SW, Rodríguez-Gironde M, Suerink M, Aretz S, Bernstein I, Capella G, Engel C, Gomez-Garcia EB, van Hest LP, von Knebel Doeberitz M, Lagerstedt-Robinson K, Letteboer TGW, Møller P, van Os TAM, Pineda M, Rahner N, Olderode-Berends MJW, von Salomé J, Schackert HK, Spruijt L, **Steinke-Lange V**, Wagner A, Tops CMJ, Nielsen M.

Cancer Epidemiol Biomarkers Prev. 1. März 2019.

### **Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes.**

Vos JR, Giepmans L, Röhl C, Geverink N, Hoogerbrugge N, **ERN GENTURIS.**

Fam Cancer. 2019;18(2):281–4.

### **Peutz-Jeghers-Syndrom**

**Steinke-Lange V, Holinski-Feder E**

TZM-News 1/2019, ISSN: 1437-8019

## **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 Dec 1;143(11):2800-2813. doi: 10.1002/ijc.31725.

### **Li-Fraumeni-Syndrom**

**Steinke-Lange V, Holinski-Feder E**

TZM-News 2/2018, ISSN: ISSN: 1437-8019

### **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**, Möslin 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, German HNPCC Consortium, the Dutch Lynch Syndrome Collaborative Group, and the Finnish Lynch Syndrome Registry.

Gastroenterology. 2018 Nov;155(5):1400-1409.e2. doi: 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, Ortmann O, Aretz S, Mallmann P, Kurzeder C, Hagen V, van Oorschot B, Höcht S, Feyer P, Egerer G, Friedrich M, Cremer W, Prott FJ, Horn LC, 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, Henscher 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 Nov;78(11):1089-1109. doi: 10.1055/a-0715-2964.

### **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, Prott FJ, Horn LC, 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, Henscher 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 Oct;78(10):949-971. doi: 10.1055/a-0713-1218.

### **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, Olderode-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 Oct 10;36(29):2961-2968. doi: 10.1200/JCO.2018.78.4777.

### **Das Cowden-Syndrom**

**Steinke-Lange V, Holinski-Feder E**

TZM-News 3-4/2018, ISSN: ISSN: 1437-8019

### **Haplotype analysis suggest that the MLH1 c.2059C > T mutation is a Swedish founder mutation.**

von Salomé J, Liu T, Keihäs M, **Morak M, Holinski-Feder E**, Berry IR, Moilanen JS, Baert-Desurmont S, Lindblom A, Lagerstedt-Robinson K.

Fam Cancer. 2018 Okt. doi: 10.1007/s10689-017-0067-x.

### **Comprehensive analysis of the MLH1 promoter region in 480 patients with colorectal cancer and 1150 controls reveals new variants including one with a heritable constitutional MLH1 epimutation.**

**Morak M**, Ibsler A, Keller G, Jessen E, **Laner A**, Gonzales-Fassrainer D, **Locher M**, Massdorf T, Nissen AM, Benet-Pagès A, **Holinski-Feder E**.

J Med Genet. 2018 Feb 22. pii: jmedgenet-2017-104744. doi: 10.1136/jmedgenet-2017-104744.

### **Identification of genetic variants for clinical management of familial colorectal tumors.**

Dominguez-Valentin M, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, **Morak M, Holinski-Feder E**, Martins A, Møller P, Hovig E.

BMC Med Genet. 2018 Feb 20;19(1):26. doi: 10.1186/s12881-018-0533-9.

### **Genetic variants of prospectively demonstrated phenocopies in BRCA1/2 kindreds.**

Dominguez-Valentin M, Evans DGR, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, **Morak M, Holinski-Feder E**, Martins A, Møller P, Hovig E.

Hered Cancer Clin Pract. 2018 Jan 15;16:4. doi: 10.1186/s13053-018-0086-0.

### **Role of germline aberrations affecting CTNNA1, MAP3K6 and MYD88 in gastric cancer susceptibility.**

Weren RDA, van der Post RS, Vogelaar IP, van Krieken JH, Spruijt L, Lubinski J, Jakubowska A, Teodorczyk U, Aalfs CM, van Hest LP, Oliveira C, Kamping EJ, Schackert HK, Ranzani GN, Gómez García EB, Hes FJ, **Holinski-Feder E**, Genuardi M, Ausems MGEM, Sijmons RH, Wagner A, van der Kolk LE, Cats A, Bjørnevoll I, Hoogerbrugge N, Ligtenberg MJL.

J Med Genet. 2018 Jan 12. pii: jmedgenet-2017-104962. doi: 10.1136/jmedgenet-2017-104962.

#### Potentially pathogenic germline CHEK2 c.319+2T>A among multiple early-onset cancer families.

Dominguez-Valentin M, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, **Morak M**, **Holinski-Feder E**, Martins A, Møller P, Hovig E.

Fam Cancer. 2018 Jan.

## 2017

#### Colorectal cancer incidence in *path\_MLH1* carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report.

Seppälä T, Pylvänäinen K, Evans DG, Järvinen H, Renkonen-Sinisalo L, Bernstein I, **Holinski-Feder E**, Sala P, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Jenkins M, Genuardi M, Green K, Laloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, **Morak M**, Frayling IM, Plazzer JP, Sampson JR, Capella G, Möslin G, Mecklin JP, Møller P; Mallorca Group.

Hered Cancer Clin Pract. 2017 Oct 10;15:18.

#### Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing.

Vogelaar IP, van der Post RS, van Krieken JHJ, Spruijt L, van Zelst-Stams WA, Kets CM, Lubinski J, Jakubowska A, Teodorczyk U, Aalfs CM, van Hest LP, Pinheiro H, Oliveira C, Jhangiani SN, Muzny DM, Gibbs RA, Lupski JR, de Ligt J, Vissers LELM, Hoischen A, Gilissen C, van de Vorst M, Goeman JJ, Schackert HK, Ranzani GN, Molinaro V, Gómez García EB, Hes FJ, **Holinski-Feder E**, Genuardi M, Ausems MGEM, Sijmons RH, Wagner A, van der Kolk LE, Bjørnevoll I, Høberg-Vetti H, van Kessel AG, Kuiper RP, Ligtenberg MJL, Hoogerbrugge N.

Eur J Hum Genet. 2017 Sep 6.

#### Cancer risk and survival in *path\_MMR* carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database.

Møller P, Seppälä TT, Bernstein I, **Holinski-Feder E**, Sala P, Gareth Evans D, Lindblom A, Macrae F, Blanco I, Sijmons RH, Jeffries J, Vasen HFA, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen JT, Jenkins MA, Green K, Laloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, **Morak M**, Renkonen-Sinisalo L, Valentin MD, Frayling IM, Plazzer JP, Pylvänäinen K, Genuardi M, Mecklin JP, Moeslein G, Sampson JR, Capella G; Mallorca Group.

Gut. 2017 Jul 28.

#### Elucidating the molecular basis of *msh2*-deficient tumors by combined germline and somatic analysis.

Vargas-Parra GM, González-Acosta M, Thompson BA, Gómez C, Fernández A, Dámaso E, Pons T, **Morak M**, Del Valle J, Iglesias S, Velasco À, Solanes A, Sanjuan X, Padilla N, de la Cruz X, Valencia A, **Holinski-Feder E**, Brunet J, Feliubadaló L, Lázaro C, Navarro M, Pineda M, Capellá G.

Int J Cancer. 2017 Jun 2.

#### Loss of MSH2 and MSH6 due to heterozygous germline defects in MSH3 and MSH6.

**Morak M**, Käsbauer S, Kerscher M, Laner A, Nissen AM, Benet-Pagès A, Schackert HK, Keller G, Massdorf T, **Holinski-Feder E**.

Fam Cancer. 2017 May 20.

#### Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database.

Møller P, Seppälä T, Bernstein I, **Holinski-Feder E**, Sala P, Evans DG, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Green K, Laloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, **Morak M**, Renkonen-Sinisalo L, Frayling IM, Plazzer JP, Pylvanainen K, Sampson JR, Capella G, Mecklin JP, Möslein G; Mallorca Group (<http://mallorca-group.eu>).  
Gut. 2017 Mar;66(3):464-472.

## 2016

### Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer.

Rump A, Benet-Pages A, Schubert S, Kuhlmann JD, Janavičius R, Macháčková E, Foretová L, Kleibl Z, Lhota F, Zemankova P, Betcheva-Krajcir E, Mackenroth L, Hackmann K, Lehmann J, Nissen A, DiDonato N, Opitz R, Thiele H, Kast K, Wimberger P, **Holinski-Feder E**, Emmert S, Schröck E, Klink B. PLoS Genet. 2016 Aug 9;12(8):e1006248.

### Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis.

Adam R, Spier I, Zhao B, Kloth M, Marquez J, Hinrichsen I, Kirfel J, Tafazzoli A, Horpaopan S, Uhlhaas S, Stienen D, Friedrichs N, Altmüller J, **Laner A**, Holzapfel S, Peters S, Kayser K, Thiele H, **Holinski-Feder E**, Marra G, Kristiansen G, Nöthen MM, Büttner R, Möslein G, Betz RC, Brieger A, Lifton RP, Aretz S.  
Am J Hum Genet. 2016 Aug 4;99(2):337-51.

### Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database.

Møller P, Seppälä T, Bernstein I, **Holinski-Feder E**, Sala P, Evans DG, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Jenkins M, Green K, Laloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, **Morak M**, Renkonen-Sinisalo L, Frayling IM, Plazzer JP, Pylvanainen K, Genuardi M, Mecklin JP, Möslein G, Sampson JR, Capella G; Mallorca Group (<http://mallorca-group.org>).  
Gut. 2016 Jun 3. pii: gutjnl-2016-311403.

### Exome sequencing identifies potential novel candidate genes in patients with unexplained colorectal adenomatous polyposis.

Spier I, Kerick M, Drichel D, Horpaopan S, Altmüller J, **Laner A**, Holzapfel S, Peters S, Adam R, Zhao B, Becker T, Lifton RP, **Holinski-Feder E**, Perner S, Thiele H, Nöthen MM, Hoffmann P, Timmermann B, Schweiger MR, Aretz S.  
Fam Cancer. 2016 Apr;15(2):281-8.

### Low-level APC mutational mosaicism is the underlying cause in a substantial fraction of unexplained colorectal adenomatous polyposis cases.

Spier I, Drichel D, Kerick M, Kirfel J, Horpaopan S, **Laner A**, Holzapfel S, Peters S, Adam R, Zhao B, Becker T, Lifton RP, Perner S, Hoffmann P, Kristiansen G, Timmermann B, Nöthen MM, **Holinski-Feder E**, Schweiger MR, Aretz S.  
J Med Genet. 2016 Mar;53(3):172-9.