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Characterization of candidate genes in unexplained polyposis and colorectal cancer

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List of publications

Mismatch repair deficiency and MUTYH variants in small intestine-neuroendocrine tumors.

Helderman NC, **Elsayed FA**, van Wezel T, Terlouw D, Langers AMJ, van Egmond D, Kiliç G, Hristova H, Farina Sarasqueta A, Morreau H, Nielsen M, Suerink M; PALGA-group collaborators. *Human pathology*. 2022 Jul;125:11-17. doi: 10.1016/j.humpath.2022.04.003.

Use of sanger and next-generation sequencing to screen for mosaic and intronic APC variants in unexplained colorectal polyposis patients.

Elsayed FA, Tops CMJ, Nielsen M, Morreau H, Hes FJ, van Wezel T. *Familial Cancer*. 2022 Jan; 21(1):79-83. doi: 10.1007/s10689-021-00236-2.

Monoallelic NTHL1 loss-of-function variants and risk of polyposis and colorectal cancer.

Elsayed FA*, Grolleman JE*, Ragunathan A*; NTHL1 study group; Buchanan DD, van Wezel T, de Voer RM. *Gastroenterology*. 2020 Dec;159(6):2241-2243.e6. doi: 10.1053/j.gastro.2020.08.042.

Low frequency of POLD1 and POLE exonuclease domain variants in patients with multiple colorectal polyps.

Elsayed FA, Tops CMJ, Nielsen M, Ruano D, Vasen HFA, Morreau H, Hes FJ, van Wezel T. *Molecular Genetics & Genomic Medicine*. 2019 Apr;7(4):e00603. doi: 10.1002/mgg3.603.

Mutational signature analysis reveals NTHL1 deficiency to cause a multi-tumor phenotype.

Grolleman JE*, de Voer RM*, **Elsayed FA***, Nielsen M*, Weren RDA*, Palles C, Ligtenberg MJL, Vos JR, Ten Broeke SW, de Miranda NFCC, Kuiper RA, Kamping EJ, Jansen EAM, Vink-Börger ME, Popp I, Lang A, Spier I, Hüneburg R, James PA, Li N, Staninova M, Lindsay H, Cockburn D, Spasic-Boskovic O, Clendenning M, Sweet K, Capellá G, Sjursen W, Høberg-Vetti H, Jongmans MC, Neveling K, Geurts van Kessel A, Morreau H, Hes FJ, Sijmons RH, Schackert HK, Ruiz-Ponte C, Dymerska D, Lubinski J, Rivera B, Foulkes WD, Tomlinson IP, Valle L, Buchanan DD, Kenwick S, Adlard J, Dimovski AJ, Campbell IG, Aretz S, Schindler D, van Wezel T, Hoogerbrugge N, Kuiper RP. *Cancer Cell*. 2019 Feb 11;35(2):256-266.e5. doi: 10.1016/j.ccell.2018.12.011. *These authors contributed equally.

SNP association study in PMS2-associated Lynch syndrome.

Ten Broeke SW, **Elsayed FA**, Pagan L, Olderode-Berends MJW, Garcia EG, Gille HJP, van Hest LP, Letteboer TGW, van der Kolk LE, Mensenkamp AR, van Os TA, Spruijt L, Redeker BJW, Suerink M, Vos YJ, Wagner A, Wijnen JT, Steyerberg EW, Tops CMJ, van Wezel T, Nielsen M. *Familial Cancer*. 2018 Oct;17(4):507-515. doi: 10.1007/s10689-017-0061-3.

Evidence for genetic association between chromosome 1q loci and predisposition to colorectal neoplasia.

Schubert SA, Ruano D, **Elsayed FA**, Boot A, Crobach S, Sarasqueta AF, Wolffenbuttel B, van der Klauw MM, Oosting J, Tops CM, van Eijk R, Vasen HF, Vossen RH, Nielsen M, Castellví-Bel S, Ruiz-Ponte C, Tomlinson I, Dunlop MG, Vodicka P, Wijnen JT, Hes FJ, Morreau H, de Miranda NF, Sijmons RH, van Wezel T. *British Journal of Cancer*. 2017 Sep 5;117(6):1215-1223. doi: 10.1038/bjc.2017.240.

Germline variants in POLE are associated with early onset mismatch repair deficient colorectal cancer.

Elsayed FA, Kets CM, Ruano D, van den Akker B, Mensenkamp AR, Schruppf M, Nielsen M, Wijnen JT, Tops CM, Ligtenberg MJ, Vasen HF, Hes FJ, Morreau H, van Wezel T. *European Journal of Human Genetics*. 2015 Aug;23(8):1080-4. doi: 10.1038/ejhg.2014.242.

Curriculum vitae

Fadwa was born in Khartoum, Sudan. She obtained the degree of bachelor of science (honours) from Faculty of Science, University of Khartoum, Sudan. After which she started working as a teaching assistant in Department of Biology and Biotechnology, Faculty of Science and Technology, Al Neelain University, Sudan. While working there she obtained her master degree from Faculty of Science, University of Khartoum, after which she was promoted to lecturer in the Department of Biology and Biotechnology, Faculty of Science and Technology, Al Neelain University. To further build her career she came to the Leiden University Medical Center, Department of Pathology in 2012 and she worked as researcher under supervision of Dr. Tom van Wezel and Dr. Frederik Hes in a collaborative project between the Departments of Pathology and Clinical Genetics. The project aimed at identification of new genes predisposing for polyposis and colorectal cancer. In 2015 she continued working on this project as a PhD candidate in the Department of Pathology and in collaboration with the Department of Clinical Genetics under supervision of Dr. Tom van Wezel, Dr. Maartje Nielsen and Prof. dr. Hans Morreau. The results of this PhD research are presented in this thesis. During her PhD she also worked on other projects not included in this thesis such as a SNP association studies in PMS2-associated Lynch syndrome and in MAP patients, and the role of digenic inheritance of *NTHL1* and *MUTYH* in predisposition colorectal cancer.

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