СПИСЪК

на цитирания на научни публикации на д-р Елица Бечева - Крайчир

ORCiD 0009-0009-8339-5637 / Web of Science Researcher ID: KLC-9167-2024 / Scopus ID 56662187800

СПИСЪК

на цитирания в SCOPUS и Web of Sciences на научни публикации
на д-р Елица Бечева - Крайчир

№	Цитирана публикация	Цитираща публикация
1	**Novel *PRPS1* gain-of-function mutation in a patient with congenital hyperuricemia and facial anomalies**. Porrmann J., **Betcheva-Krajcir E**., Di Donato N., Kahlert A.-K., Schallner J., Rump A., Schrock E., Dobritzsch, D., Roelofsen, J., Tzschach A. (**2017**) American Journal of Medical Genetics, Part A, 173 (10) , pp. 2736-2742.	1. Dewulf, J.P., Marie, S., Nassogne, M.-C. Disorders of purine biosynthesis metabolism (2022) Molecular Genetics and Metabolism, 136 (3), pp. 190-198 2. Marie, S., Dewulf, J.P., Nassogne, M.-C. Disorders of purine and pyrimidine metabolism (2022) Inborn Metabolic Diseases: Diagnosis and Treatment, pp. 587-614 3. Ugbogu, E.A., Schweizer, L.M., Schweizer, M. Contribution of Model Organisms to Investigating the Far-Reaching Consequences of PRPP Metabolism on Human Health and Well-Being (2022) Cells, 11 (12), art. no. 1909 4. Lenherr, N., Christodoulou, J., Duley, J., Dobritzsch, D., Fairbanks, L., Datta, A.N., Filges, I., Gürtler, N., Roelofsen, J., van Kuilenburg, A.B.P., Kemper, C., West, E.E., Szinnai, G., Huemer, M. Co-therapy with S-adenosylmethionine and nicotinamide riboside improves t-cell survival and function in Arts Syndrome (PRPS1 deficiency) (2021) Molecular Genetics and Metabolism Reports, 26, art. no. 100709 5. Puusepp, S., Reinson, K., Pajusalu, S., van Kuilenburg, A.B.P., Dobritzsch, D., Roelofsen, J., Stenzel, W., Õunap, K. Atypical presentation of Arts syndrome due to a novel hemizygous loss-of-function variant in the PRPS1 gene (2020) Molecular Genetics and Metabolism Reports, 25, art. no. 100677 6. Mercati, O., Abi Warde, M.-T., Lina-Granade, G., Rio, M., Heide, S., de Lonlay, P., Ceballos-Picot, I., Robert, M.P., Couloigner, V., Beltrand, J., Boddaert, N., Rodriguez, D., Rubinato, E., Lapierre, J.-M., Merlette, C., Sanquer, S., Rötig, A., Prokisch, H., Lyonnet, S., Loundon, N., Kaplan, J., Bonnefont, J.-P., Munnich, A., Besmond, C., Jonard, L., Marlin, S. PRPS1 loss-of-function variants, from isolated hearing loss to severe congenital encephalopathy: New cases and literature review (2020) European Journal of Medical Genetics, 63 (11), art. no. 104033 7. Yang, B.-Y., Yu, H.-X., Min, J., Song, X.-X. A novel mutation in gene of PRPS1 in a young Chinese woman with X-linked gout: a case report and review of the literature (2020) Clinical Rheumatology, 39 (3), pp. 949-956 8. van der Wijst, J., Belge, H., Bindels, R.J.M., Devuyst, O. Learning physiology from inherited kidney disorders (2019) Physiological Reviews, 99 (3), pp. 1575-1653 9. Saugstad, O.D. 50 Years Ago in THE JOURNAL OF PEDIATRICS: A New Disorder of Purine Metabolism with Behavioral Manifestations (2019) Journal of Pediatrics, 204, p. 88. 10. Zikánova, M., Wahezi, D., Hay, A., Stiburková, B., Pitts, C., Mušálková, D., Škopová, V., Barešová, V., Součková, O., Hodaňová, K., Živná, M., Stránecký, V., Hartmannová, H., Hnízda, A., Bleyer, A.J., Kmoch, S. Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females (2018) Rheumatology (United Kingdom), 57 (7), pp. 1180-1185 11. Zhu, W., Deng, Y., Zhou, X. Multiple membrane transporters and some immune regulatory genes are major genetic factors to gout (2018) Open Rheumatology Journal, 12 (1), pp. 94-113
2	**Skewed X-inactivation in a family with *DLG3*-associated X-linked intellectual disability**. Gieldon L., Mackenroth L., **Betcheva-Krajcir E**., Rump A., Beck-Wodl S., Schallner J., Di Donato N., Schröck, E., Tzschach A. (**2017**) American Journal of Medical Genetics, Part A, 173 (9) , pp. 2545-2550.	1. Kunde SA, Schmerl B, von Sivers J, Ahmadyar E, Gupta T, Rademacher N, Zieger HL, Shoichet SA. JNK activity modulates postsynaptic scaffold protein SAP102 and kainate receptor dynamics in dendritic spines. J Biol Chem. 2024 Apr 4;300(5):107263. doi: 10.1016/j.jbc.2024.107263. Epub ahead of print. PMID: 38582451; PMCID: PMC11081805. 2. Huyghebaert, J., Mateiu, L., Elinck, E., Van Rossem, K.E., Christiaenssen, B., D'Incal, C.P., McCormack, M.K., Lazzarini, A., Vandeweyer, G., Kooy, R.F. Identification of a DLG3 stop mutation in the MRX20 family (2024) European Journal of Human Genetics 3. Kharrat, M., Issa, A.B., Tlili, A., Jallouli, O., Alila-Fersi, O., Maalej, M., Chouchen, J., Ghouylia, Y., Kamoun, F., Triki, C., Fakhfakh, F. A Novel Mutation in the MAP7D3 Gene in Two Siblings with Severe

		Intellectual Disability and Autistic Traits: Concurrent Assessment of BDNF Functional Polymorphism, X-Inactivation and Oxidative Stress to Explain Disease Severity (2023) Journal of Molecular Neuroscience, 73 (9-10), pp. 853-864 4. Tolmacheva, E.N., Fonova, E.A., Lebedev, I.N. X-Linked CNV in Pathogenetics of Intellectual Disability (2022) Russian Journal of Genetics, 58 (10), pp. 1193-1207. 5. Jia, H., Hongjie, Z., Jiahuan, H., Xi, L., Xingxing, L., Hongdan, W., Congmin, L., Yue, W., Hongyan, L. Pedigree analysis and prenatal diagnosis of intellectual disability caused by synonymous mutations of p.S434S in DLG3 gene (2022) Chinese Journal of Perinatal Medicine, 25 (1), pp. 42-47 6. Pereira, G., Dória, S. X-chromosome inactivation: implications in human disease (2021) Journal of Genetics, 100 (2), art. no. 63 7. Zhang, X., Qiu, W., Liu, H., Ye, X., Sun, Y., Fan, Y., Yu, Y. RT-PCR analysis of mRNA revealed the splice-altering effect of rare intronic variants in monogenic disorders (2020) Annals of Human Genetics, 84 (6), pp. 456-462 8. Matis, T., Michaud, V., Van-Gils, J., Raclet, V., Plaisant, C., Fergelot, P., Lasseaux, E., Arveiler, B., Trimouille, A. Triple diagnosis of Wiedemann-Steiner, Waardenburg and DLG3-related intellectual disability association found by WES: A case report (2020) Journal of Gene Medicine, 22 (8), art. no. e3197 9. Tolmacheva, E.N., Kashevarova, A.A., Nazarenko, L.P., Minaycheva, L.I., Skryabin, N.A., Lopatkina, M.E., Nikitina, T.V., Sazhenova, E.A., Belyaeva, E.O., Fonova, E.A., Salyukova, O.A., Tarabykin, V.S., Lebedev, I.N. Delineation of clinical manifestations of the inherited xq24 microdeletion segregating with sxci in mothers: Two novel cases with distinct phenotypes ranging from ube2a deficiency syndrome to recurrent pregnancy loss (2020) Cytogenetic and Genome Research, 160 (5), pp. 245-254 10. Carmignac, V., Nambot, S., Lehalle, D., Callier, P., Moortgat, S., Benoit, V., Ghoumid, J., Delobel, B., Smol, T., Thuillier, C., Zordan, C., Naudion, S., Bienvenu, T., Touraine, R., Ramond, F., Zweier, C., Reis, A., Kraus, C., Nizon, M., Cogné, B., Verloes, A., Tran Mau-Them, F., Sorlin, A., Jouan, T., Duffourd, Y., Tisserant, E., Philippe, C., Vitobello, A., Thevenon, J., Faivre, L., Thauvin-Robinet, C. Further delineation of the female phenotype with KDM5C disease causing variants: 19 new individuals and review of the literature (2020) Clinical Genetics, 98 (1), pp. 43-55 11. Sandestig, A., Green, A., Aronsson, J., Ellnebo, K., Stefanova, M. A Novel DLG3 Mutation Expanding the Phenotype of X-Linked Intellectual Disability Caused by DLG3 Nonsense Variants (2019) Molecular Syndromology, 10 (5), pp. 281-285. 12. Liu, M., Wang, Y., Yang, S., Wei, H., Tuo, M., Chang, F., Wang, Y. Single nucleotide polymorphism array analysis uncovers a large, novel duplication in Xq13.1 in a floppy infant syndrome patient (2019) International Journal of Developmental Neuroscience, 74, pp. 56-60 13. Pozzi, D., Menna, E., Canzi, A., Desiato, G., Mantovani, C., Matteoli, M. The communication between the immune and nervous systems: The role of IL-1β in synaptopathies (2018) Frontiers in Molecular Neuroscience, 11, art. no. 111 14. Brand BA, Blesson AE, Smith-Hicks CL. The Impact of X-Chromosome Inactivation on Phenotypic Expression of X-Linked Neurodevelopmental Disorders. Brain Sci. 2021 Jul 9;11(7):904. doi: 10.3390/brainsci11070904. PMID: 34356138; PMCID: PMC8305405.
3	**Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH**. Hackmann K., Kuhlee F., **Betcheva-Krajcir E**., Kahlert A.-K., Mackenroth L., Klink B., Di Donato N., Tzschach, A., Kast, K., Wimberger, P., Schrock, E., Rump A. (**2016**) Breast Cancer Research and Treatment, 159 (3) , pp. 585-590.	1. Pereira, D.M., Fernandes, J.C. The power of genomics, metabolomics, and other omics for target identification and validation (2021) Principles of Translational Science in Medicine: From Bench to Bedside, Third Edition, pp. 33-48 2. Lepkes, L., Kayali, M., Blümcke, B., Weber, J., Suszynska, M., Schmidt, S., Borde, J., Klonowska, K., Wappenschmidt, B., Hauke, J., Kozlowski, P., Schmutzler, R.K., Hahnen, E., Ernst, C. Performance of in silico prediction tools for the detection of germline copy number variations in cancer predisposition genes in 4208 female index patients with familial breast and ovarian cancer

		(2021) Cancers, 13 (1), art. no. 118, pp. 1-12 3. Schubert, S., van Luttikhuizen, J.L., Auber, B., Schmidt, G., Hofmann, W., Penkert, J., Davenport, C.F., Hille-Betz, U., Wendeburg, L., Bublitz, J., Tauscher, M., Hackmann, K., Schröck, E., Scholz, C., Wallaschek, H., Schlegelberger, B., Illig, T., Steinemann, D. The identification of pathogenic variants in BRCA1/2 negative, high risk, hereditary breast and/or ovarian cancer patients: High frequency of FANCM pathogenic variants (2019) International Journal of Cancer, 144 (11), pp. 2683-2694. 4. Zakrzewski, F., Gieldon, L., Rump, A., Seifert, M., Grützmann, K., Krüger, A., Loos, S., Zeugner, S., Hackmann, K., Porrmann, J., Wagner, J., Kast, K., Wimberger, P., Baretton, G., Schröck, E., Aust, D., Klink, B. Targeted capture-based NGS is superior to multiplex PCR-based NGS for hereditary BRCA1 and BRCA2 gene analysis in FFPE tumor samples (2019) BMC Cancer, 19 (1), art. no. 396 5. Rizza, R., Hackmann, K., Paris, I., Minucci, A., De Leo, R., Schrock, E., Urbani, A., Capoluongo, E., Gelli, G., Concolino, P. Novel BRCA1 Large Genomic Rearrangements in Italian Breast/Ovarian Cancer Patients (2019) Molecular Diagnosis and Therapy, 23 (1), pp. 121-126 6. Ding, M., Gao, J., Ling, C., Gao, L. CnnCNV: A Sensitive and Efficient Method for Detecting Copy Number Variation based on Convolutional Neural Networks (2019) Proceedings - 2018 IEEE International Conference on Bioinformatics and Biomedicine, BIBM 2018, art. no. 8621321, pp. 2744-2746. 7. Zheng, M., Hu, Y., Gou, R., Wang, J., Nie, X., Li, X., Liu, Q., Liu, J., Lin, B. Integrated multi-omics analysis of genomics, epigenomics, and transcriptomics in ovarian carcinoma (2019) Aging, 11 (12), pp. 4198-4215. 8. Gieldon, L., William, D., Hackmann, K., Jahn, W., Jahn, A., Wagner, J., Rump, A., Bechmann, N., Nölting, S., Knösel, T., Gudziol, V., Constantinescu, G., Masjkur, J., Beuschlein, F., Timmers, H.J.L.M., Canu, L., Pacak, K., Robledo, M., Aust, D., Schröck, E., Eisenhofer, G., Richter, S., Klink, B. Optimizing genetic workup in pheochromocytoma and paraganglioma by integrating diagnostic and research approaches (2019) Cancers, 11 (6), art. no. 809 9. Penkert, J., Schmidt, G., Hofmann, W., Schubert, S., Schieck, M., Auber, B., Ripperger, T., Hackmann, K., Sturm, M., Prokisch, H., Hille-Betz, U., Mark, D., Illig, T., Schlegelberger, B., Steinemann, D. Breast cancer patients suggestive of Li-Fraumeni syndrome: Mutational spectrum, candidate genes, and unexplained heredity (2018) Breast Cancer Research, 20 (1), art. no. 87 10. Liu, W., Gao, J. An integrated method of detecting copy number variation based on sequence assembly (2018) Lecture Notes in Computer Science (including subseries Lecture Notes in Artificial Intelligence and Lecture Notes in Bioinformatics), 10954 LNCS, pp. 589-594 11. Concolino, P., Rizza, R., Hackmann, K., Minucci, A., Scaglione, G.L., De Bonis, M., Costella, A., Zuppi, C., Schrock, E., Capoluongo, E. Identification and Characterization of a New BRCA2 Rearrangement in an Italian Family with Hereditary Breast and Ovarian Cancer Syndrome (2017) Molecular Diagnosis and Therapy, 21 (5), pp. 539-545 12. Concolino, P., Rizza, R., Hackmann, K., Paris, I., Minucci, A., De Paolis, E., Scambia, G., Zuppi, C., Schrock, E., Capoluongo, E. Characterization of a new BRCA1 rearrangement in an Italian woman with hereditary breast and ovarian cancer syndrome (2017) Breast Cancer Research and Treatment, 164 (2), pp. 497-503 13. Lantieri, F., Malacarne, M., Gimelli, S., Santamaria, G., Coviello, D., Ceccherini, I. Custom array comparative genomic hybridization: The importance of DNA quality, an expert eye, and variant validation (2017) International Journal of Molecular Sciences, 18 (3), art. no. 609 14. Schubert, S., Ripperger, T., Rood, M., Petkidis, A., Hofmann, W., Frye-Boukhriss, H., Tauscher, M., Auber, B., Hille-Betz, U., Illig, T., Schlegelberger, B., Steinemann, D. GT198 (PSMC3IP) germline variants in early-onset breast cancer patients from hereditary breast and ovarian cancer families (2017) Genes and Cancer, 8 (1-2), pp.

		472-483 15. Sun, G.-F., Ding, H. Cloning of rat C-sis gene and construction of its eukaryotic expression vector (2016) Chinese Journal of Tissue Engineering Research, 20 (49), pp. 7418-7424
4	**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 J.D., Janavicius R., Machackova E., Foretova 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. (**2016**) PLoS Genetics, 12 (8) , art. no. e1006248	1. Tuncer, S.B., Celik, B., Erciyas, S.K., Erdogan, O.S., Gültaslar, B.K., Odemis, D.A., Avsar, M., Sen, F., Saip, P.M., Yazici, H. Germline mutational variants of Turkish ovarian cancer patients suspected of Hereditary Breast and Ovarian Cancer (HBOC) by next-generation sequencing (2024) Pathology Research and Practice, 254, art. no. 155075 2. Pasqui, A., Boddi, A., Campanacci, D.A., Scoccianti, G., Bernini, A., Grasso, D., Gambale, E., Scolari, F., Palchetti, I., Palomba, A., Fancelli, S., Caliman, E., Antonuzzo, L., Pillozzi, S. Alteration of the Nucleotide Excision Repair (NER) Pathway in Soft Tissue Sarcoma (2022) International Journal of Molecular Sciences, 23 (15), art. no. 8360 3. Dumont, M., Weber-Lassalle, N., Joly-Beauparlant, C., Ernst, C., Droit, A., Feng, B.-J., Dubois, S., Collin-Deschesnes, A.-C., Soucy, P., Vallée, M., Fournier, F., Lemaçon, A., Adank, M.A., Allen, J., Altmüller, J., Arnold, N., Ausems, M.G.E.M., Berutti, R., Bolla, M.K., Bull, S., Carvalho, S., Cornelissen, S., Dufault, M.R., Dunning, A.M., Engel, C., Gehrig, A., Geurts-Giele, W.R.R., Gieger, C., Green, J., Hackmann, K., Helmy, M., Hentschel, J., Hogervorst, F.B.L., Hollestelle, A., Hooning, M.J., Horváth, J., Ikram, M.A., Kaulfuß, S., Keeman, R., Kuang, D., Luccarini, C., Maier, W., Martens, J.W.M., Niederacher, D., Nürnberg, P., Ott, C.-E., Peters, A., Pharoah, P.D.P., Ramirez, A., Ramser, J., Riedel-Heller, S., Schmidt, G., Shah, M., Scherer, M., Stäbler, A., Strom, T.M., Sutter, C., Thiele, H., van Asperen, C.J., van der Kolk, L., van der Luijt, R.B., Volk, A.E., Wagner, M., Waisfisz, Q., Wang, Q., Wang-Gohrke, S., Weber, B.H.F., Devilee, P., Tavtigian, S., Bader, G.D., Meindl, A., Goldgar, D.E., Andrulis, I.L., Schmutzler, R.K., Easton, D.F., Schmidt, M.K., Hahnen, E., Simard, J. Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry (2022) Cancers, 14 (14), art. no. 3363 4. Baszuk, P., Stadnik, P., Marciniak, W., Derkacz, R., Jakubowska, A., Cybulski, C., Huzarski, T., Gronwald, J., Dębniak, T., Białkowska, K., Pietrzak, S., Kładny, J., Scott, R.J., Lubiński, J., Lener, M.R. Low blood-as levels and selected genotypes appears to be promising biomarkers for occurrence of colorectal cancer in women (2021) Biomedicines, 9 (9), art. no. 1105 5. Wieme, G., Kral, J., Rosseel, T., Zemankova, P., Parton, B., Vocka, M., Van Heetvelde, M., Kleiblova, P., Blaumeiser, B., Soukupova, J., van den Ende, J., Nehasil, P., Tejpar, S., Borecka, M., García, E.B.G., Blok, M.J., Safarikova, M., Kalousova, M., Geboes, K., De Putter, R., Poppe, B., De Leeneer, K., Kleibl, Z., Janatova, M., Claes, K.B.M. Prevalence of germline pathogenic variants in cancer predisposing genes in czech and belgian pancreatic cancer patients (2021) Cancers, 13 (17), art. no. 4430 6. Hait, A.S., Thomsen, M.M., Larsen, S.M., Helleberg, M., Mardahl, M., Barfod, T.S., Christiansen, M., Brandt, C., Mogensen, T.H. Whole-Exome Sequencing of Patients With Recurrent HSV-2 Lymphocytic Mollaret Meningitis (2021) Journal of Infectious Diseases, 223 (10), pp. 1776-1786 7. Doddato, G., Valentino, F., Giliberti, A., Papa, F.T., Tita, R., Bruno, L.P., Resciniti, S., Fallerini, C., Benetti, E., Palmieri, M., Mencarelli, M.A., Fabbiani, A., Bruttini, M., Orrico, A., Baldassarri, M., Fava, F., Lopergolo, D., Lo Rizzo, C., Lamacchia, V., Mannucci, S., Pinto, A.M., Currò, A., Mancini, V., Mari, F., Renieri, A., Ariani, F. Whole Exome Sequencing in BRCA1-2 Candidate Families: The Contribution of Other Cancer Susceptibility Genes (2021) Frontiers in Oncology, 11, art. no. 649435 8. Tao, J.-J., Yang, C., Li, J.-F., Pan, H.-Q., Chen, W.-J., Yang, B. Mutation in the ERCC2 gene identified in a Chinese trichothiodystrophy patient (2021) Journal of Dermatology, 48 (5),

		pp. e203-e204 9. Topka, S., Steinsnyder, Z., Ravichandran, V., Tkachuk, K., Kemel, Y., Bandlamudi, C., Madsen, M.W., Furberg, H., Ouerfelli, O., Rudin, C.M., Iyer, G., Lipkin, S.M., Mukherjee, S., Solit, D.B., Berger, M.F., Bajorin, D.F., Rosenberg, J.E., Taylor, B.S., de Stanchina, E., Vijai, J., Offit, K. Targeting germline- And tumor-associated nucleotide excision repair defects in cancer (2021) Clinical Cancer Research, 27 (7), pp. 1997-2010 10. Kim, G., Kim, J., Cha, H., Park, W.-Y., Ahn, J.S., Ahn, M.-J., Park, K., Park, Y.-J., Choi, J.Y., Lee, K.-H., Lee, S.-H., Moon, S.H. Metabolic radiogenomics in lung cancer: associations between FDG PET image features and oncogenic signaling pathway alterations (2020) Scientific Reports, 10 (1), art. no. 13231 11. Chernock, R.D., Rivera, B., Borrelli, N., Hill, D.A., Fahiminiya, S., Shah, T., Chong, A.-S., Aqil, B., Mehrad, M., Giordano, T.J., Sheridan, R., Rutter, M.M., Dehner, L.P., Foulkes, W.D., Nikiforov, Y.E. Poorly differentiated thyroid carcinoma of childhood and adolescence: a distinct entity characterized by DICER1 mutations (2020) Modern Pathology, 33 (7), pp. 1264-1274 12. Fankhauser, M., Bechmann, N., Lauseker, M., Goncalves, J., Favier, J., Klink, B., William, D., Gieldon, L., Maurer, J., Spöttl, G., Rank, P., Knösel, T., Orth, M., Ziegler, C.G., Prada, E.T.A., Rubinstein, G., Fassnacht, M., Spitzweg, C., Grossman, A.B., Pacak, K., Beuschlein, F., Bornstein, S.R., Eisenhofer, G., Auernhammer, C.J., Reincke, M., Nölting, S. Synergistic Highly Potent Targeted Drug Combinations in Different Pheochromocytoma Models Including Human Tumor Cultures (2019) Endocrinology (United States), 160 (11), pp. 2600-2617 13. Zakrzewski, F., Gieldon, L., Rump, A., Seifert, M., Grützmann, K., Krüger, A., Loos, S., Zeugner, S., Hackmann, K., Porrmann, J., Wagner, J., Kast, K., Wimberger, P., Baretton, G., Schröck, E., Aust, D., Klink, B. Targeted capture-based NGS is superior to multiplex PCR-based NGS for hereditary BRCA1 and BRCA2 gene analysis in FFPE tumor samples (2019) BMC Cancer, 19 (1), art. no. 396, 14. Díaz-Gay, M., Franch-Expósito, S., Arnau-Collell, C., Park, S., Supek, F., Muñoz, J., Bonjoch, L., Gratacós-Mulleras, A., Sánchez-Rojas, P.A., Esteban-Jurado, C., Ocaña, T., Cuatrecasas, M., Vila-Casadesús, M., Lozano, J.J., Parra, G., Laurie, S., Beltran, S., Castells, A., Bujanda, L., Cubiella, J., Balaguer, F., Castellví-Bel, S. Integrated analysis of germline and tumor DNA identifies new candidate genes involved in familial colorectal cancer (2019) Cancers, 11 (3), art. no. 362 15. Richter, S., Gieldon, L., Pang, Y., Peitzsch, M., Huynh, T., Leton, R., Viana, B., Ercolino, T., Mangelis, A., Rapizzi, E., Menschikowski, M., Aust, D., Kroiss, M., Beuschlein, F., Gudziol, V., Timmers, H.J., Lenders, J., Mannelli, M., Cascon, A., Pacak, K., Robledo, M., Eisenhofer, G., Klink, B. Metabolome-guided genomics to identify pathogenic variants in isocitrate dehydrogenase, fumarate hydratase, and succinate dehydrogenase genes in pheochromocytoma and paraganglioma (2019) Genetics in Medicine, 21 (3), pp. 705-717 16. Koudová, M., Puchmajerová, A. Risks of solid tumors in heterozygous carriers of recessive syndromes (2019) Klinicka Onkologie, 32, pp. 2S14-2S23 17. Boldrin, E., Malacrida, S., Rumiato, E., Battaglia, G., Ruol, A., Amadori, A., Saggioro, D. Association between ERCC1rs3212986 and ERCC2/XPDrs1799793 and OS in Patients with Advanced Esophageal Cancer (2019) Frontiers in Oncology, 9 (FEB), art. no. 85 18. Abdullah, M.-I., Muhammad, N.A.N. Prediction of colorectal cancer driver genes from patients' genome data (2018) Sains Malaysiana, 47 (12), pp. 3095-3105 19. Aravind Kumar, M., Naushad, S.M., Narasimgu, N., Nagaraju Naik, S., Kadali, S., Shanker, U., Lakshmi Narasu, M. Whole exome sequencing of breast cancer (TNBC) cases from India: association of MSH6 and BRIP1 variants with TNBC risk and oxidative DNA damage (2018) Molecular Biology Reports, 45 (5), pp. 1413-1419 20. Yehia, L., Ni, Y., Sesock, K., Niazi, F., Fletcher, B., Chen, H.J., LaFramboise, T., Eng, C. Unexpected cancer-predisposition gene

		variants in Cowden syndrome and Bannayan-Riley-Ruvalcaba syndrome patients without underlying germline PTEN mutations (2018) PLoS Genetics, 14 (4), art. no. e1007352 21. Kim, C., Yang, J., Jeong, S.-H., Kim, H., Park, G.-H., Shin, H.B., Ro, M., Kim, K.-Y., Park, Y., Kim, K.P., Kwack, K. Yeast-based assays for characterization of the functional effects of single nucleotide polymorphisms in human DNA repair genes (2018) PLoS ONE, 13 (3), art. no. e0193823 22. Gieldon, L., Masjkur, J.R., Richter, S., Därr, R., Lahera, M., Aust, D., Zeugner, S., Rump, A., Hackmann, K., Tzschach, A., Januszewicz, A., Prejbisz, A., Eisenhofer, G., Schrock, E., Robledo, M., Klink, B. Next-generation panel sequencing identifies NF1 germline mutations in three patients with pheochromocytoma but no clinical diagnosis of neurofibromatosis type 1 (2018) European Journal of Endocrinology, 178 (2), pp. K1-K9 23. Zhang, W., Zhang, Z. Associations between XRCC2 rs3218536 and ERCC2 rs13181 polymorphisms and ovarian cancer (2016) Oncotarget, 7 (52), pp. 86621-86630
5	**HBOC multi-gene panel testing: comparison of two sequencing centers**. Schroeder C., Faust U., Sturm M., Hackmann K., Grundmann K., Harmuth F., Bosse K., Kehrer, M., Benkert, T., Klink, B., Mackenroth, L., **Betcheva-Krajcir, E**., Wimberger, P., Kast, K., Heilig, M., Nguyen, H., Riess, O., Schröck, E., Bauer, P., Rump, A. (**2015**) Breast Cancer Research and Treatment, 152 (1) , pp. 129-136.	1. Zang, B., Helms, M., Besch, L., Kalmbach, N., Stegen, S., Blohmer, J.-U., Speiser, D. Interdisciplinary risk counseling for hereditary breast and ovarian cancer: real-world data from a specialized center (2023) Archives of Gynecology and Obstetrics, 307 (5), pp. 1585-1592 2. Goidescu, I.G., Nemeti, G., Surcel, M., Caracostea, G., Florian, A.R., Cruciat, G., Staicu, A., Muresan, D., Goidescu, C., Pintican, R., Eniu, D.T. Spectrum of High-Risk Mutations among Breast Cancer Patients Referred for Multigene Panel Testing in a Romanian Population (2023) Cancers, 15 (6), art. no. 1895 3. Zhunussova, G., Omarbayeva, N., Kaidarova, D., Abdikerim, S., Mit, N., Kisselev, I., Yergali, K., Zhunussova, A., Goncharova, T., Abdrakhmanova, A., Djansugurova, L. Determination of genetic predisposition to early breast cancer in women of Kazakh ethnicity (2023) Oncotarget, 14, pp. 860-877 4. Castillo-Guardiola, V., Rosado-Jiménez, L., Sarabia-Meseguer, M.D., Marín-Vera, M., Macías-Cerrolaza, J.A., García-Hernández, R., Zafra-Poves, M., Sánchez-Henarejos, P., Moreno-Locubiche, M.Á., Cuevas-Tortosa, E., Arnaldos-Carrillo, M., Ayala de la Peña, F., Alonso-Romero, J.L., Noguera-Velasco, J.A., Ruiz-Espejo, F. Next step in molecular genetics of hereditary breast/ovarian cancer: Multigene panel testing in clinical actionably genes and prioritization algorithms in the study of variants of uncertain significance (2022) European Journal of Medical Genetics, 65 (4), art. no. 104468 5. Lesueur, F., Easton, D.F., Renault, A.-L., Tavtigian, S.V., Bernstein, J.L., Kote-Jarai, Z., Eeles, R.A., Plaseska-Karanfia, D., Feliubadaló, L., Moles-Fernández, A., Santamariña-Pena, M., Sánchez, A.T., López-Novo, A., Porras, L.-M., Blanco, A., Capellá, G., de la Hoya, M., Molina, I.J., Osorio, A., Pineda, M., Rueda, D., de la Cruz, X., Diez, O., Ruiz-Ponte, C., Gutiérrez-Enríquez, S., Vega, A., Lázaro, C., Arun, B., Herold, N., Versmold, B., Schmutzler, R.K., Nguyen-Dumont, T., Southey, M.C., Dorling, L., Dunning, A.M., Ghiorzo, P., Dalmasso, B.S., Cavaciuti, E., Le Gal, D., Roberts, N.J., Dominguez-Valentin, M., Rookus, M., Taylor, A.M.R., Goldstein, A.M., Goldgar, D.E., Couch, F., Kraft, P., Weitzel, J., Nathanson, K., Domchek, S., LaDuca, H., Stoppa-Lyonnet, D., Andrieu, N. First international workshop of the ATM and cancer risk group (4-5 December 2019) (2022) Familial Cancer, 21 (2), pp. 211-227 6. Rasool, R., Ullah, I., Mubeen, B., Alshehri, S., Imam, S.S., Ghoneim, M.M., Alzarea, S.I., Al-Abbasi, F.A., Murtaza, B.N., Kazmi, I., Nadeem, M.S. Theranostic Interpolation of Genomic Instability in Breast Cancer (2022) International Journal of Molecular Sciences, 23 (3), art. no. 1861 7. Pereira, M.A., de Oliveira Marinho, F.L., Leite, L.R., Alvim, L.B., Malta, F.S.V., Marques, F.K., de Lima, A.B. Breast Cancer and Next-Generation Sequencing: Towards Clinical Relevance and Future (2022) Breast Cancer: From Bench to Personalized Medicine, pp. 477-518 8. Tomioka, K., Miyamoto, T., Akutsu, S.N., Yanagihara, H., Fujita, K., Royba, E., Tauchi, H., Yamamoto, T., Koh, I., Hirata, E., Kudo, Y., Kobayashi, M., Okada, S., Matsuura, S. NBS1 I171V variant underlies

		individual differences in chromosomal radiosensitivity within human populations (2021) Scientific Reports, 11 (1), art. no. 19661 9. Rajan, A., Nadhan, R., Latha, N.R., Krishnan, N., Warrier, A.V., Srinivas, P. Deregulated estrogen receptor signaling and DNA damage response in breast tumorigenesis (2021) Biochimica et Biophysica Acta - Reviews on Cancer, 1875 (1), art. no. 188482 10. Dhooge, M., Baert-Desurmont, S., Corsini, C., Caron, O., Andrieu, N., Berthet, P., Bonadona, V., Cohen-Haguenauer, O., De Pauw, A., Delnatte, C., Dussart, S., Lasset, C., Leroux, D., Maugard, C., Moretta-Serra, J., Popovici, C., Buecher, B., Colas, C., Noguès, C. National recommendations of the French Genetics and Cancer Group - Unicancer on the modalities of multi-genes panel analyses in hereditary predispositions to tumors of the digestive tract (2020) European Journal of Medical Genetics, 63 (12), art. no. 104080 11. Kwong, A., Shin, V.Y., Chen, J., Cheuk, I.W.Y., Ho, C.Y.S., Au, C.H., Chan, K.K.L., Ngan, H.Y.S., Chan, T.L., Ford, J.M., Ma, E.S.K. Germline Mutation in 1338 BRCA-Negative Chinese Hereditary Breast and/or Ovarian Cancer Patients: Clinical Testing with a Multigene Test Panel (2020) Journal of Molecular Diagnostics, 22 (4), pp. 544-554 12. Da Costa E Silva Carvalho, S., Cury, N.M., Brotto, D.B., De Araujo, L.F., Rosa, R.C.A., Texeira, L.A., Plaça, J.R., Marques, A.A., Peronni, K.C., Ruy, P.D.C., Molfetta, G.A., Moriguti, J.C., Carraro, D.M., Palmero, E.I., Ashton-Prolla, P., De Faria Ferraz, V.E., Silva, W.A. Germline variants in DNA repair genes associated with hereditary breast and ovarian cancer syndrome: Analysis of a 21 gene panel in the Brazilian population (2020) BMC Medical Genomics, 13 (1), art. no. 21 13. Flaum, N., Crosbie, E.J., Edmondson, R.J., Smith, M.J., Evans, D.G. Epithelial ovarian cancer risk: A review of the current genetic landscape (2020) Clinical Genetics, 97 (1), pp. 54-63 14. Ryu, J.M., Nam, S.J., Kim, S.W., Lee, J.E., Chae, B.J., Lee, S.K., Yu, J. Prognosis of BRCA1/2-negative breast cancer patients with HBOC risk factors compared with sporadic breast cancer patients without HBOC risk factors (2019) Japanese Journal of Clinical Oncology, 50 (2), pp. 104-113 15. Dennis, K., Silvia, A.-B., Katharina, S., Paranchai, B., Markus, Z., Dedes Konstantin, J., Pascal, J., Daniel, F., Anita, R. Prevalence of genetic susceptibility for breast and ovarian cancer in a non-cancer related study population: Secondary germline findings from a Swiss single centre cohort (2019) Swiss Medical Weekly, 149 (33-34), art. no. w20092 16. Jacobs, M.F., Dust, H., Koeppe, E., Wong, S., Mulholland, M., Choi, E.-Y., Appelman, H., Stoffel, E.M. Outcomes of Endoscopic Surveillance in Individuals With Genetic Predisposition to Hereditary Diffuse Gastric Cancer (2019) Gastroenterology, 157 (1), pp. 87-96 17. Suszynska, M., Kluzniak, W., Wokolorczyk, D., Jakubowska, A., Huzarski, T., Gronwald, J., Debniak, T., Szwiec, M., Ratajska, M., Klonowska, K., Narod, S., Bogdanova, N., Dörk, T., Lubinski, J., Cybulski, C., Kozlowski, P. Bard1 is a low/moderate breast cancer risk gene: Evidence based on an association study of the central european p.q564x recurrent mutation (2019) Cancers, 11 (6), art. no. 740 18. Schubert, S., van Luttikhuizen, J.L., Auber, B., Schmidt, G., Hofmann, W., Penkert, J., Davenport, C.F., Hille-Betz, U., Wendeburg, L., Bublitz, J., Tauscher, M., Hackmann, K., Schröck, E., Scholz, C., Wallaschek, H., Schlegelberger, B., Illig, T., Steinemann, D. The identification of pathogenic variants in BRCA1/2 negative, high risk, hereditary breast and/or ovarian cancer patients: High frequency of FANCM pathogenic variants (2019) International Journal of Cancer, 144 (11), pp. 2683-2694 19. Zakrzewski, F., Gieldon, L., Rump, A., Seifert, M., Grützmann, K., Krüger, A., Loos, S., Zeugner, S., Hackmann, K., Porrmann, J., Wagner, J., Kast, K., Wimberger, P., Baretton, G., Schröck, E., Aust, D., Klink, B. Targeted capture-based NGS is superior to multiplex PCR-based NGS for hereditary BRCA1 and BRCA2 gene analysis in FFPE tumor samples (2019) BMC Cancer, 19 (1), art. no. 396 20. Catana, A., Apostu, A.P., Antemie, R.-G. Multi gene panel testing for hereditary breast cancer - Is it ready to be used? (2019) Medicine and

Pharmacy Reports, 92 (3), pp. 220-225
21. Gieldon, L., William, D., Hackmann, K., Jahn, W., Jahn, A., Wagner, J., Rump, A., Bechmann, N., Nölting, S., Knösel, T., Gudziol, V., Constantinescu, G., Masjkur, J., Beuschlein, F., Timmers, H.J.L.M., Canu, L., Pacak, K., Robledo, M., Aust, D., Schröck, E., Eisenhofer, G., Richter, S., Klink, B. Optimizing genetic workup in pheochromocytoma and paraganglioma by integrating diagnostic and research approaches (2019) Cancers, 11 (6), art. no. 809
22. Bonache, S., Esteban, I., Moles-Fernández, A., Tenés, A., Duran-Lozano, L., Montalban, G., Bach, V., Carrasco, E., Gadea, N., López-Fernández, A., Torres-Esquius, S., Mancuso, F., Caratú, G., Vivancos, A., Tuset, N., Balmaña, J., Gutiérrez-Enríquez, S., Diez, O. Multigene panel testing beyond BRCA1/2 in breast/ovarian cancer Spanish families and clinical actionability of findings (2018) Journal of Cancer Research and Clinical Oncology, 144 (12), pp. 2495-2513
23. Fortuno, C., James, P.A., Spurdle, A.B. Current review of TP53 pathogenic germline variants in breast cancer patients outside Li-Fraumeni syndrome (2018) Human Mutation, 39 (12), pp. 1764-1773
24. Urban, R.Q., Velásquez, C.E.D., Gitler, R., Castillo, M.P.R., Toporek, M.S., Morales, A.F., García, O.M., Esquivel, L.G., Mejía, G.T., Dean, M., Enciso, I.D., López, H.O.D., León, F.R., Jan, V., Barrientos, V.H.G., Flores, P.R., Silva, P.K.E., Cruz, J.H.S., Gregorio, H.M., Jiménez, E.A.R., Cruz, L.E.R., Catalá, C.F.M., Gómez, R.M.Á., Ontiveros, V.F., Herrera, L.A., Romieu, I., Terrazas, L.I., Chirino, Y.I., Frecha, C., Oliver, J., Perdomo, S., Paniagua, F.V. Comprehensive analysis of germline variants in mexican patients with hereditary breast and ovarian cancer susceptibility (2018) Cancers, 10 (10), art. no. 361
25. Maksimenko, J., Irmejs, A., Trofimovičs, G., Berziņa, D., Skuja, E., Purkalne, G., Miklaševičs, E., Gardovskis, J. High frequency of pathogenic non-founder germline mutations in BRCA1 and BRCA2 in families with breast and ovarian cancer in a founder population (2018) Hereditary Cancer in Clinical Practice, 16 (1), art. no. 12
26. Low, S.-K., Zembutsu, H., Nakamura, Y. Breast cancer: The translation of big genomic data to cancer precision medicine (2018) Cancer Science, 109 (3), pp. 497-506
27. Gieldon, L., Masjkur, J.R., Richter, S., Därr, R., Lahera, M., Aust, D., Zeugner, S., Rump, A., Hackmann, K., Tzschach, A., Januszewicz, A., Prejbisz, A., Eisenhofer, G., Schrock, E., Robledo, M., Klink, B. Next-generation panel sequencing identifies NF1 germline mutations in three patients with pheochromocytoma but no clinical diagnosis of neurofibromatosis type 1 (2018) European Journal of Endocrinology, 178 (2), pp. K1-K9
28. O'Donnell, M., Axilbund, J., Euhus, D.M. Breast cancer genetics: Syndromes, genes, pathology, counseling, testing, and treatment (2018) The Breast: Comprehensive Management of Benign and Malignant Diseases, pp. 237-249.e5.
29. Oda, K., Tanikawa, M., Sone, K., Mori-Uchino, M., Osuga, Y., Fujii, T. Recent advances in targeting DNA repair pathways for the treatment of ovarian cancer and their clinical relevance (2017) International Journal of Clinical Oncology, 22 (4), pp. 611-618
30. Suresh, P.S., Venkatesh, T., Tsutsumi, R., Shetty, A. Next-generation sequencing for endocrine cancers: Recent advances and challenges (2017) Tumor Biology, 39 (5)
31. Kamps, R., Brandão, R.D., van den Bosch, B.J., Paulussen, A.D.C., Xanthoulea, S., Blok, M.J., Romano, A. Next-generation sequencing in oncology: Genetic diagnosis, risk prediction and cancer classification (2017) International Journal of Molecular Sciences, 18 (2), art. no. 308
32. Eliade, M., Skrzypski, J., Baurand, A., Jacquot, C., Bertolone, G., Loustalot, C., Coutant, C., Guy, F., Fumoleau, P., Duffourd, Y., Arnould, L., Delignette, A., Padéano, M.-M., Lepage, C., Raichon-Patru, G., Boudrant, A., Bône-Lépinoy, M.-C., Villing, A.-L., Charpin, A., Peignaux, K., Chevrier, S., Vegran, F., Ghiringhelli, F., Boidot, R., Sevenet, N., Lizard, S., Faivre, L. The transfer of multigene panel testing for hereditary breast and ovarian cancer to healthcare: What are the implications for the management of patients and families? (2017) Oncotarget, 8 (2), pp. 1957-1971
33. Lhota, F., Zemankova, P., Kleiblova, P., Soukupova, J., Vocka, M., Stranecky, V., Janatova, M., Hartmannova, H., Hodanova, K., Kmoch,

		S., Kleibl, Z. Hereditary truncating mutations of DNA repair and other genes in BRCA1/BRCA2/PALB2-negatively tested breast cancer patients (2016) Clinical Genetics, 90 (4), pp. 324-333 34. Hackmann, K., Kuhlee, F., Betcheva-Krajcir, E., Kahlert, A.-K., Mackenroth, L., Klink, B., Di Donato, N., Tzschach, A., Kast, K., Wimberger, P., Schrock, E., Rump, A. Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH (2016) Breast Cancer Research and Treatment, 159 (3), pp. 585-590 35. Paolillo, C., Londin, E., Fortina, P. Next generation sequencing in cancer: opportunities and challenges for precision cancer medicine (2016) Scandinavian Journal of Clinical and Laboratory Investigation, 76, pp. S84-S91. 36. Pistorius, S., Klink, B., Pablik, J., Rump, A., Aust, D., Garzarolli, M., Schröck, E., Schackert, H.K. An unusual case of Cowden syndrome associated with ganglioneuromatous polyposis (2016) Hereditary Cancer in Clinical Practice, 14 (1), art. no. 11 37. Song, J., Eichstaedt, C.A., Viales, R.R., Benjamin, N., Harutyunova, S., Fischer, C., Grünig, E., Hinderhofer, K. Identification of genetic defects in pulmonary arterial hypertension by a new gene panel diagnostic tool (2016) Clinical Science, 130 (22), pp. 2043-2052 38. Soukupová, J., Zemánková, P., Kleiblová, P., Janatová, M., Kleibl, Z. CZECANCA: Czech cancer panel for clinical application - Design and optimization of the targeted sequencing panel for the identification of cancer susceptibility in high-risk individuals from the Czech Republic (2016) Klinicka Onkologie, 29, pp. S46-S54. 39. Lerner-Ellis, J., Khalouei, S., Sopik, V., Narod, S.A. Genetic risk assessment and prevention: The role of genetic testing panels in breast cancer (2015) Expert Review of Anticancer Therapy, 15 (11), pp. 1315-1326
6	**Whole-genome-wide association study in the Bulgarian population reveals *HHAT* as schizophrenia susceptibility gene**. **Betcheva E.T.**, Yosifova A.G., Mushiroda T., Kubo M., Takahashi A., Karachanak S.K., Zaharieva I.T., Hadjidekova, S., Dimova, I., Vazharova, R., Stoyanov, D., Nakamura Y. (**2013**) Psychiatric Genetics, 23 (1), pp. 11-19.	1. Wang, J., Liu, J., Li, S., Li, X., Yang, J., Dang, X., Mu, C., Li, Y., Li, K., Li, J., Chen, R., Liu, Y., Huang, D., Zhang, Z., Luo, X.-J. Genetic regulatory and biological implications of the 10q24.32 schizophrenia risk locus (2023) Brain, 146 (4), pp. 1403-1419 2. Abdelfattah, A.M., Abuelezz, S.A., Hendawy, N., Negm, E.A., Nawishy, S.A.E.K., Khalil, A.M.M. Sonic hedgehog pathway as a new target of atypical antipsychotics: Revisiting of amisulpride and aripiprazole effects in a rat model of schizophrenia (2023) Life Sciences, 316, art. no. 121366, 3. Bokulic, E., Medenica, T., Sedmak, G. Transcriptional Profile of the Developing Subthalamic Nucleus (2022) eNeuro, 9 (5), art. no. ENEURO.0193-22.2022 4. Pol-Fuster, J., Cañellas, F., Ruiz-Guerra, L., Medina-Dols, A., Bisbal-Carrió, B., Asensio, V., Ortega-Vila, B., Marzese, D., Vidal, C., Santos, C., Lladó, J., Olmos, G., Heine-Suñer, D., Strauch, K., Flaquer, A., Vives-Bauzà, C. Familial Psychosis Associated With a Missense Mutation at MACF1 Gene Combined With the Rare Duplications DUP3p26.3 and DUP16q23.3, Affecting the CNTN6 and CDH13 Genes (2021) Frontiers in Genetics, 12, art. no. 622886 5. Ivanov, H.Y., Velinov, B., Kyosovska, G., Grigorova, D., Shopov, P. Exploring Pharmacogenetic Variation in a Bulgarian Psychiatric Cohort (2021) Folia Medica, 63 (3), pp. 365-371 6. Mueller, T.M., Meador-Woodruff, J.H. Post-translational protein modifications in schizophrenia (2020) npj Schizophrenia, 6 (1), art. no. 5 7. Zhang, Z., Chen, G. A logical relationship for schizophrenia, bipolar, and major depressive disorder. Part 1: Evidence from chromosome 1 high density association screen (2020) Journal of Comparative Neurology, 528 (15), pp. 2620-2635 8. Prata, D.P., Costa-Neves, B., Cosme, G., Vassos, E. Unravelling the genetic basis of schizophrenia and bipolar disorder with GWAS: A systematic review (2019) Journal of Psychiatric Research, 114, pp. 178-207 9. Rhoades, R., Jackson, F., Teng, S. Discovery of rare variants implicated in schizophrenia using next-generation sequencing (2019) Journal of Translational Genetics and Genomics, 3, art. no. 1 10. Wang, Z., Li, P., Wu, T., Zhu, S., Deng, L., Cui, G. Axon guidance pathway genes are associated with schizophrenia risk (2018)

		Experimental and Therapeutic Medicine, 16 (6), pp. 4519-4526 11. Khan, F.F., Melton, P.E., McCarthy, N.S., Morar, B., Blangero, J., Moses, E.K., Jablensky, A. Whole genome sequencing of 91 multiplex schizophrenia families reveals increased burden of rare, exonic copy number variation in schizophrenia probands and genetic heterogeneity (2018) Schizophrenia Research, 197, pp. 337-345 12. Memi, F., Zecevic, N., Radonjić, N. Multiple roles of Sonic Hedgehog in the developing human cortex are suggested by its widespread distribution (2018) Brain Structure and Function, 223 (5), pp. 2361-2375 13. Misiewicz, Z., Hiekkalinna, T., Paunio, T., Varilo, T., Terwilliger, J.D., Partonen, T., Hovatta, I. A genome-wide screen for acrophobia susceptibility loci in a Finnish isolate (2016) Scientific Reports, 6, art. no. 39345 14. Zhang, W., Lin, J.-R., Nogales-Cadenas, R., Zhang, Q., Cai, Y., Zhang, Z.D. Prioritization of schizophrenia risk genes by a network-regularized logistic regression method (2016) Lecture Notes in Computer Science (including subseries Lecture Notes in Artificial Intelligence and Lecture Notes in Bioinformatics), 9656, pp. 434-445 15. Li, Z., Xiang, Y., Chen, J., Li, Q., Shen, J., Liu, Y., Li, W., Xing, Q., Wang, Q., Wang, L., Feng, G., He, L., Zhao, X., Shi, Y. Loci with genome-wide associations with schizophrenia in the Han Chinese population (2015) British Journal of Psychiatry, 207 (6), pp. 490-494 16. Stoyanov, D.S., Stieglitz, R.-D., Lenz, C., Borgwardt, S.J. The translational validation as novel approach to integration of neuroscience and psychiatry (2015) New Developments in Clinical Psychology Research, pp. 195-208 17. Walton, E., Turner, J.A., Ehrlich, S. Neuroimaging as a potential biomarker to optimize psychiatric research and treatment (2013) International Review of Psychiatry, 25 (5), pp. 619-631 18. Thomas, T.R., Koomar, T., Casten, L.G. et al. Clinical autism subscales have common genetic liabilities that are heritable, pleiotropic, and generalizable to the general population. Transl Psychiatry 12, 247 (2022). https://doi.org/10.1038/s41398-022-01982-2
7	**Methylenetetrahydrofolate reductase (*MTHFR*) C677T and A1298C polymorphisms and age of onset in schizophrenia: A combined analysis of independent samples**. Saetre P., Vares M., Werge T., Andreassen O.A., Arinami T., Ishiguro H., Nanko S., Tan, Ch., Han, D., Roffman, J., Muntjewerff, Jan-Willem, Jagodzinski, P., Kempisty, B., Hauser, J., Vilella, **E., Betcheva**, E., Nakamura, Y., Regland, B., Agartz, I., Hall, H., Terenius, L., Jonsson E.G. (**2011**) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 156 (2), pp. 215-224.	1. Wan, L., Wei, J. Early-onset schizophrenia: A special phenotype of the disease characterized by increased mthfr polymorphisms and aggravating symptoms (2021) Neuropsychiatric Disease and Treatment, 17, pp. 2511-2525 2. Wan, L., Li, Y., Zhou, Y., Li, R., Zheng, Y. Age Matters: an Atypical Association Between Polymorphism of MTHFR and Clinical Phenotypes in Children with Schizophrenia (2019) Journal of Molecular Neuroscience, 69 (3), pp. 485-493 3. Ai Chin, J.T., Gunasekaran, G., Damanhuri, M.H.A., Fong, C.L., Lin, L.J., Aan, G.J. Oxidative status in bipolar disorder (BD) and its correlation with age, gender and body mass index (BMI) (2019) Sains Malaysiana, 48 (5), pp. 1083-1095. 4. Wan, L., Li, Y., Zhang, Z., Sun, Z., He, Y., Li, R. Methylenetetrahydrofolate reductase and psychiatric diseases (2018) Translational Psychiatry, 8 (1), art. no. 242 5. Fond, G., Godin, O., Boyer, L., Llorca, P.-M., Andrianarisoa, M., Brunel, L., Aouizerate, B., Berna, F., Capdevielle, D., D'Amato, T., Denizot, H., Dubertret, C., Dubreucq, J., Faget, C., Gabayet, F., Mallet, J., Misdrahi, D., Passerieux, C., Rey, R., Richieri, R., Schandrin, A., Urbach, M., vidailhet, P., Leboyer, M., Bulzacka, E., Schürhoff, F., Andrianarisoa, M., Aouizerate, B., Berna, F., Blanc, O., Brunel, L., Bulzacka, E., Capdevielle, D., Chereau-Boudet, I., Chesnoy-Servanin, G., Danion, J.-M., D'Amato, T., Deloge, A., Delorme, C., Denizot, H., Dorey, J.-M., Dubertret, C., Dubreucq, J., Faget, C., Fluttaz, C., Fond, G., Fonteneau, S., Giraud-Baro, E., Hardy-Bayle, M.-C., Delphine, L., Lançon, C., Laouamri, H., Leboyer, M., Le Gloahec, T., Le Strat, Y., Llorca, P.-M., Mallet, J., Metairie, E., Offerlin-Meyer, I., Passerieux, C., Peri, P., Pires, S., Portalier, C., Rey, R., Roman, C., Schandrin, A., Schürhoff, F., Tessier, A., Tronche, A.-M., Urbach, M., Vaillant, F., Vehier, A., Vidailhet, P., Vilà, E., Yazbek, H., Zinetti-Bertschy, A. Advanced paternal age is associated with earlier schizophrenia onset in offspring. Results from the national multicentric FACE-SZ cohort

(2017) Psychiatry Research, 254, pp. 218-223.
6. Duprey, R.P. MTHFR gene polymorphism positive treatment-resistant depression: Prevalence and treatment recommendations (2016) Neuropsychiatry, 6 (2), pp. 43-46
7. Zainullina, A., Valiullina, A., Khusnutdinova, E. The role of genetic and environmental risk factors in aetiology of suicidal behaviour (2016) Behavioural Genetics for Education, pp. 205-230
8. Hu, C.-Y., Qian, Z.-Z., Gong, F.-F., Lu, S.-S., Feng, F., Wu, Y.-L., Yang, H.-Y., Sun, Y.-H. Methylenetetrahydrofolate reductase (MTHFR) polymorphism susceptibility to schizophrenia and bipolar disorder: an updated meta-analysis (2015) Journal of Neural Transmission, 122 (2), pp. 307-320
9. Liew, S.-C., Gupta, E.D. Methylenetetrahydrofolate reductase (MTHFR) C677T polymorphism: Epidemiology, metabolism and the associated diseases (2015) European Journal of Medical Genetics, 58 (1), pp. 1-10
10. Misiak, B., Frydecka, D., Slezak, R., Piotrowski, P., Kiejna, A. Elevated homocysteine level in first-episode schizophrenia patients - The relevance of family history of schizophrenia and lifetime diagnosis of cannabis abuse (2014) Metabolic Brain Disease, 29 (3), pp. 661-670
11. El-Hadidy, M.A., Abdeen, H.M., Abd El-Aziz, S.M., Al-Harrass, M. MTHFR gene polymorphism and age of onset of schizophrenia and bipolar disorder (2014) BioMed Research International, 2014, art. no. 318483
12. Zhang, J., Xiao, Y., Zhang, X.-W., Gao, Z.-Q., Han, J.-H. Relationship between methylenetetrahydrofolate reductase (MTHFR) A1298C gene polymorphism and type 2 diabetic nephropathy risk: A meta-analysis (2014) Renal Failure, 36 (6), pp. 974-978
13. Geller, V., Friger, M., Sela, B.-A., Levine, J. Elevated homocysteine level in siblings of patients with schizophrenia (2013) Psychiatry Research, 210 (3), pp. 769-772
14. Lochman, J., Plesník, J., Janout, V., Povová, J., Míšek, I., Dvořáková, D., Šerý, O. Interactive effect of MTHFR and ADRA2A gene polymorphisms on pathogenesis of schizophrenia (2013) Neuroendocrinology Letters, 34 (8), pp. 792-797
15. Sampogna, F., Camaioni, D.B., Giampetruzzi, A.R., Corona, R., Ruzzi, L., Silvestri, L., Didona, B. Chronic idiopathic acrocyanosis and methylenetetrahydrofolate reductase C677T (p.Ala222Val) and A1298C (p.Glu429Ala) polymorphisms (2013) European Journal of Dermatology, 23 (3), pp. 356-361
16. Ekerholm, M., Firus Waltersson, S., Fagerberg, T., Söderman, E., Terenius, L., Agartz, I., Jönsson, E.G., Nyman, H. Neurocognitive function in long-term treated schizophrenia: A five-year follow-up study (2012) Psychiatry Research, 200 (2-3), pp. 144-152
17. Fard-Esfahani, P., Torbati, P.M., Hashemi, Z., Fayaz, S., Golkar, M. Analysis of relation between C677T genotype in MTHFR gene and prostatic cancer in Iranian males (2012) Acta Medica Iranica, 50 (10), pp. 657-663
18. Chojnicka, I., Sobczyk-Kopcioł, A., Fudalej, M., Fudalej, S., Wojnar, M., Waśkiewicz, A., Broda, G., Strawa, K., Pawlak, A., Krajewski, P., Płoski, R. No association between MTHFR C677T polymorphism and completed suicide (2012) Gene, 511 (1), pp. 118-121.
19. Saetre, P., Grove, J., Børglum, A.D., Mors, O., Werge, T., Andreassen, O.A., Vares, M., Agartz, I., Terenius, L., Jönsson, E.G. Methylenetetrahydrofolate reductase (MTHFR) C677T polymorphism and age at onset of schizophrenia: No consistent evidence for an association in the nordic population (2012) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 159 B (8), pp. 981-986
20. Antonsen, J.H., Gonda, X., Dome, P., Rihmer, Z. Associations between season of birth and suicide: A brief review (2012) Neuropsychopharmacologia Hungarica, 14 (3), pp. 177-187.
21. Keshavan, M.S., Sanghavi, K. Psychiatric genetics research in Asia (2012) Asian Journal of Psychiatry, 5 (2), pp. 123-124.
22. Lajin, B., Alhaj Sakur, A., Michati, R., Alachkar, A. Association between MTHFR C677T and A1298C, and MTRR A66G

		polymorphisms and susceptibility to schizophrenia in a Syrian study cohort (2012) Asian Journal of Psychiatry, 5 (2), pp. 144-149 23. Cui, W.-P., Du, B., Jia, Y., Zhou, W.-H., Liu, S.-M., Cui, Y.-C., Ma, F.-Z., Luo, P., Miao, L.-N. Is C677T Polymorphism in Methylenetetrahydrofolate Reductase Gene a Risk Factor for Diabetic Nephropathy or Diabetes Mellitus in a Chinese Population? (2012) Archives of Medical Research, 43 (1), pp. 42-50
8	**Web-based software for storage, statistical processing and analysis of snp data in studies on complex disorders**. **Betcheva E**., Betchev C., Toncheva D. (**2009**) Balkan Journal of Medical Genetics, 12 (1), pp. 9-14. **ДОКТОРСКА СТЕПЕН**	1. Ispanovic-Radojkovic, V., Stancheva-Popkostadinova, V. Perspectives of intellectual disability in Serbia and Bulgaria: Epidemiology, policy and services for children and adults (2011) Current Opinion in Psychiatry, 24 (5), pp. 419-424
9	**Case-control association study of 59 candidate genes reveals the *DRD2* SNP rs6277 (C957T) as the only susceptibility factor for schizophrenia in the Bulgarian population**. **Betcheva E.T**., Mushiroda T., Takahashi A., Kubo M., Karachanak S.K., Zaharieva I.T., Vazharova R.V., Dimova, I., Milanova, V., Tolev, T., Kirov, G., Owen, M., O'Donovan, M., Kamatani, N., Nakamura, Y., Toncheva D.I. (**2009**) Journal of Human Genetics, 54 (2), pp. 98-107. **ДОКТОРСКА СТЕПЕН**	1. Kang, Y., Zhang, Y., Huang, K., Wang, Z. Association of dopamine-based genetic risk score with dynamic low-frequency fluctuations in first-episode drug-naïve schizophrenia (2023) Brain Imaging and Behavior, 17 (6), pp. 584-594 2. Frydecka, D., Hamza, E.A., Helal, A., Moustafa, A.A. Childhood traumatic events and the dopaminergic theory of psychosis: A mini-review of studies investigating gene – environment interactions (2023) Current Psychology, 42 (14), pp. 12296-12305 3. Lynham, A.J., Knott, S., Underwood, J.F.G., Hubbard, L., Agha, S.S., Bisson, J.I., Van Den Bree, M.B.M., Chawner, S.J.R.A., Craddock, N., O'Donovan, M., Jones, I.R., Kirov, G., Langley, K., Martin, J., Rice, F., Roberts, N.P., Thapar, A., Anney, R., Owen, M.J., Hall, J., Pardiñas, A.F., Walters, J.T.R. DRAGON-Data: A platform and protocol for integrating genomic and phenotypic data across large psychiatric cohorts (2023) BJPsych Open, 9 (2), art. no. e32 4. Ivanova, E. A PATIENT WITH SCHIZOPHRENIA AND DEUTERANOPIA (2023) Bulgarian Journal of Psychiatry, 8 (3), pp. 33-37 5. Marques, J.H., Talib, L.L., Hortêncio, L., Andrade, J.C., Alves, T.M., Serpa, M.H., Yamamoto, G.L., van de Bilt, M.T., Rössler, W., Gattaz, W.F., Loch, A.A. A dopamine receptor D2 genetic polymorphism associated with transition to mental disorders in a cohort of individuals with at-risk mental state for psychosis (2023) Brazilian Journal of Psychiatry, 45 (3), pp. 268-273. 6. Zhang, Y.-X., Yang, L.-P., Gai, C., Cheng, C.-C., Guo, Z.-Y., Sun, H.-M., Hu, D. Association between variants of MTHFR genes and psychiatric disorders: A meta-analysis (2022) Frontiers in Psychiatry, 13, art. no. 976428 7. Meng, X., Zheng, J.-L., Sun, M.-L., Lai, H.-Y., Wang, B.-J., Yao, J., Wang, H. Association between MTHFR (677C>T and 1298A>C) polymorphisms and psychiatric disorder: A meta-analysis (2022) PLoS ONE, 17 (7 July), art. no. e0271170 8. Xu, F.-L., Yao, J., Wang, B.-J. Association between RGS4 gene polymorphisms and schizophrenia: A protocol for systematic review and meta-analysis (2021) Medicine (United States), 100 (44), art. no. e27607 9. Habibzadeh, P., Nemati, A., Dastsooz, H., Taghipour-Sheshdeh, A., Paul, P.M., Sahraian, A., Faghihi, M.A. Investigating the association between common DRD2/ANKK1 genetic polymorphisms and schizophrenia: a meta-analysis (2021) Journal of Genetics, 100 (2), art. no. 59 10. Zhang, Y., Li, S., Li, X., Yang, Y., Li, W., Xiao, X., Li, M., Lv, L., Luo, X. Convergent lines of evidence support NOTCH4 as a schizophrenia risk gene (2021) Journal of Medical Genetics, 58 (10), pp. 666-678 11. Gomez, D., Power, C., Fujiwara, E. Neurocognitive Impairment and Associated Genetic Aspects in HIV Infection (2021) Current Topics in Behavioral Neurosciences, 50, pp. 41-76 12. Adaimé, A., Hajj, A., Hallit, S., Karam Sarkis, D. Lebanese observatory of pathogenic agents (lopa-study) a 2 year-surveillance prospective study (2021) Journal Medical Libanais, 69 (1), pp. 126-

		133 13. Korovaitseva, G.I., Gabaeva, M.V., Golimbet, V.E. Studies of the Association of Genetic Polymorphism C677T in the Methylenetetrahydrofolate Reductase Gene with Symptom Severity in Schizophrenia Patients (2021) Neuroscience and Behavioral Physiology, 51 (1), pp. 7-11. 14. Adaimé, A., Hajj, A., Hallit, S., Karam Sarkis, D. Lebanese observatory of pathogenic agents (lopa-study) a 2 year-surveillance prospective study (2020) Journal Medical Libanais, 68 (3), pp. 126-133 15. Korovaitseva, G.I., Gabaeva, M.V., Golimbet, V.E. The study of the association between the c677t polymorphism of the methylenetetrahydrofolate reductase gene and severity of symptoms in patients with schizophrenia (2020) Zhurnal Nevrologii i Psihiatrii imeni S.S. Korsakova, 120 (3), pp. 48-52 16. Faron-Górecka, A., Kuśmider, M., Solich, J., Górecki, A., Dziedzicka-Wasylewska, M. Genetic variants in dopamine receptors influence on heterodimerization in the context of antipsychotic drug action (2020) Progress in Molecular Biology and Translational Science, 169, pp. 279-296 17. Tsermpini, E.-E., Skokou, M., Kordou, Z., Patrinos, G.P. Genomic basis of psychiatric illnesses and response to psychiatric drug treatment modalities (2019) Applied Genomics and Public Health, pp. 73-110 18. Mohameda, Z.I., Teeb, S.F., Tanga, P.Y. Association of functional polymorphisms in 3-Untranslated regions of COMT, DISC1, and DTNBP1 with schizophrenia: A meta-analysis (2018) Psychiatric Genetics, 28 (6), pp. 110-119 19. Ma, J.-H., Sun, X.-Y., Guo, T.-J., Barot, E., Wang, D.-F., Yan, L.-L., Ni, D.-W., Huang, N.-H., Xie, Q., Zeng, J., Ou-Yang, L., Liu, Y.-Q., Lu, Q.-B. Association on DISC1 SNPs with schizophrenia risk: A meta-analysis (2018) Psychiatry Research, 270, pp. 306-309 20. Ma, J., Wang, B., Gao, X., Wu, H., Wang, D., Li, N., Tan, J., Wang, J., Yan, L. A comparative study of the typical toxic metals in serum by patients of schizophrenia and healthy controls in China (2018) Psychiatry Research, 269, pp. 558-564 21. Mohammadi, A., Rashidi, E., Amooeian, V.G. Brain, blood, cerebrospinal fluid, and serum biomarkers in schizophrenia (2018) Psychiatry Research, 265, pp. 25-38 22. Li, M.D. Tobacco smoking addiction: Epidemiology, genetics, mechanisms, and treatment (2018) Tobacco Smoking Addiction: Epidemiology, Genetics, Mechanisms, and Treatment, pp. 1-365. 23. Barbutti, I., Machado-Neto, J.A., Arfelli, V.C., de Melo Campos, P., Traina, F., Saad, S.T.O., Archangelo, L.F. The U2AF homology motif kinase 1 (UHMK1) is upregulated upon hematopoietic cell differentiation (2018) Biochimica et Biophysica Acta - Molecular Basis of Disease, 1864 (3), pp. 959-966. 24. Wang, H.-Y., Liu, Y., Yan, J.-W., Hu, X.-L., Zhu, D.-M., Xu, X.-T., Li, X.-S. Gene polymorphisms of DISC1 is associated with schizophrenia: Evidence from a meta-analysis (2018) Progress in Neuro-Psychopharmacology and Biological Psychiatry, 81, pp. 64-73 25. Arfelli, V.C., Archangelo, L.F. UHMK1 (U2AF homology motif kinase 1) (2018) Atlas of Genetics and Cytogenetics in Oncology and Haematology, 22 (8), pp. 328-335 26. Han, J., Li, Y., Wang, X. Potential link between genetic polymorphisms of catechol-O-methyltransferase and dopamine receptors and treatment efficacy of risperidone on schizophrenia (2017) Neuropsychiatric Disease and Treatment, 13, pp. 2935-2943 27. Chen, Y.-T., Lin, C.-H., Huang, C.-H., Liang, W.-M., Lane, H.-Y. PICK1 Genetic Variation and Cognitive Function in Patients with Schizophrenia (2017) Scientific Reports, 7 (1), art. no. 1889 28. Bi, Y., Huang, X., Niu, W., Chen, S., Wu, X., Cao, Y., Zhang, R., Yang, F., Wang, L., Li, W., Xu, Y., He, L., Yu, T., He, G. No association between SLC6A2, SLC6A3, DRD2 polymorphisms and schizophrenia in the Han Chinese population (2017) Psychiatry Research, 253, pp. 398-400 29. Saini, S.M., Mancuso, S.G., Mostaid, M.S., Liu, C., Pantelis, C.,

		Everall, I.P., Bousman, C.A. Meta-analysis supports GWAS-implicated link between GRM3 and schizophrenia risk (2017) Translational Psychiatry, 7 (8), art. no. e1196 30. Smith, C.T., Dang, L.C., Buckholtz, J.W., Tetreault, A.M., Cowan, R.L., Kessler, R.M., Zald, D.H. The impact of common dopamine D2 receptor gene polymorphisms on D2/3 receptor availability: C957T as a key determinant in putamen and ventral striatum (2017) Translational Psychiatry, 7 (4), art. no. e1091 31. Rai, V., Yadav, U., Kumar, P., Yadav, S.K., Gupta, S. Methylenetetrahydrofolate reductase A1298C genetic variant & risk of schizophrenia: A meta-analysis (2017) Indian Journal of Medical Research, 145 (April), pp. 437-447 32. Al-Eitan, L., Al-Habahbeh, S., Alkhatib, R. Genetic association analysis of ERBB4 polymorphisms with the risk of schizophrenia susceptibility in a Jordanian population of Arab descent (2017) Turkish Journal of Medical Sciences, 47 (2), pp. 542-553 33. He, H., Wu, H., Yang, L., Gao, F., Fan, Y., Feng, J., Ma, X. Associations between dopamine D2 receptor gene polymorphisms and schizophrenia risk: A PRISMA compliant meta-analysis (2016) Neuropsychiatric Disease and Treatment, 12, pp. 3129-3144 34. González-Castro, T.B., Hernández-Díaz, Y., Juárez-Rojop, I.E., López-Narváez, M.L., Tovilla-Zárate, C.A., Genis-Mendoza, A., Alpuin-Reyes, M. The role of C957T, TaqI and Ser311Cys polymorphisms of the DRD2 gene in schizophrenia: Systematic review and meta-analysis (2016) Behavioral and Brain Functions, 12 (1), art. no. 29 35. Goto, Y., Lee, Y.-A., Yamaguchi, Y., Jas, E. Biological mechanisms underlying evolutionary origins of psychotic and mood disorders (2016) Neuroscience Research, 111, pp. 13-24. 36. Lawford, B.R., Barnes, M., Morris, C.P., Noble, E.P., Nyst, P., Heslop, K., Young, R.M., Voisey, J., Connor, J.P. Dopamine 2 receptor genes are associated with raised blood glucose in schizophrenia (2016) Canadian Journal of Psychiatry, 61 (5), pp. 291-297 37. Gyurkovics, M., Kotyuk, E., Katonai, E.R., Horvath, E.Z., Vereczkei, A., Szekely, A. Individual differences in flow proneness are linked to a dopamine D2 receptor gene variant (2016) Consciousness and Cognition, 42, pp. 1-8 38. Yadav, U., Kumar, P., Gupta, S., Rai, V. Role of MTHFR C677T gene polymorphism in the susceptibility of schizophrenia: An updated meta-analysis (2016) Asian Journal of Psychiatry, 20, pp. 41-51 39. Numata, S., Kinoshita, M., Tajima, A., Nishi, A., Imoto, I., Ohmori, T. Evaluation of an association between plasma total homocysteine and schizophrenia by a Mendelian randomization analysis (2015) BMC Medical Genetics, 16 (1), art. no. 54 40. Ramsay, H., Barnett, J.H., Miettunen, J., Mukkala, S., Mäki, P., Liuhanen, J., Murray, G.K., Jarvelin, M.-R., Ollila, H., Paunio, T., Veijola, J. Association between dopamine receptor D2 (DRD2) variations rs6277 and rs1800497 and cognitive performance according to risk type for psychosis: A nested case control study in a Finnish population sample (2015) PLoS ONE, 10 (6), art. no. e0127602 41. Zhang, X.Y., Chen, D.-C., Tan, Y.-L., Tan, S.-P., Luo, X., Zuo, L., Rao, W., Yu, Q., Kou, C., Allen, M., Correll, C.U., Wu, J., Soares, J.C. A functional polymorphism in the interleukin-1beta and severity of nicotine dependence in male schizophrenia: A case-control study (2015) Journal of Psychiatric Research, 64, pp. 51-58. 42. Su, L., Long, J., Pan, R., Xie, X., Mao, X., Zhou, Y., Chen, Q., Wei, B. Influence of NRGN rs12807809 polymorphism on symptom severity in individuals with schizophrenia in the Han population but not the Zhuang population of south China (2015) Acta Neuropsychiatrica, 27 (4), pp. 221-227 43. Ma, Y., Yuan, W., Jiang, X., Cui, W.-Y., Li, M.D. Updated Findings of the Association and Functional Studies of DRD2/ANKK1 Variants with Addictions (2015) Molecular Neurobiology, 51 (1), pp. 281-299 44. Hu, C.-Y., Qian, Z.-Z., Gong, F.-F., Lu, S.-S., Feng, F., Wu, Y.-L.,

		Yang, H.-Y., Sun, Y.-H. Methylenetetrahydrofolate reductase (MTHFR) polymorphism susceptibility to schizophrenia and bipolar disorder: an updated meta-analysis (2015) Journal of Neural Transmission, 122 (2), pp. 307-320 45. Zhang, B., Fan, Q.R., Li, W.H., Lu, N., Fu, D.K., Kang, Y.J., Wang, N., Li, T., Wen, X.P., Li, D.X. Association of the NOTCH4 Gene Polymorphism rs204993 with Schizophrenia in the Chinese Han Population (2015) BioMed Research International, 2015, art. no. 408096 46. Göllner, T., Fieder, M. Selection in the dopamine receptor 2 gene: A candidate SNP study (2015) PeerJ, 2015 (8), art. no. e1149 47. Rukova, B., Staneva, R., Hadjidekova, S., Stamenov, G., Milanova, V., Toncheva, D. Genome-wide methylation profiling of schizophrenia (2014) Balkan Journal of Medical Genetics, 17 (2), pp. 15-24 48. Liu, L., Fan, D., Ding, N., Hu, Y., Cai, G., Wang, L., Xin, L., Xia, Q., Li, X., Xu, S., Xu, J., Yang, X., Zou, Y., Pan, F. The relationship between DRD2 gene polymorphisms (C957T and C939T) and schizophrenia: A meta-analysis (2014) Neuroscience Letters, 583, pp. 43-48. 49. Rutten, B.P.F., Van Os, J. Epigenetic epidemiology of psychiatric disorders (2014) Epigenetic Epidemiology, pp. 343-376. 50. Walton, E., Geisler, D., Lee, P.H., Hass, J., Turner, J.A., Liu, J., Sponheim, S.R., White, T., Wassink, T.H., Roessner, V., Gollub, R.L., Calhoun, V.D., Ehrlich, S. Prefrontal inefficiency is associated with polygenic risk for schizophrenia (2014) Schizophrenia Bulletin, 40 (6), pp. 1263-1271. 51. Zhang, C., Zhang, J., Fan, J., Cheng, W., Du, Y., Yu, S., Fang, Y. Identification of ANKK1 rs1800497 variant in schizophrenia: New data and meta-analysis (2014) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 165 (7), pp. 564-571 52. Bruenig, D., White, M.J., Young, R.M., Voisey, J. Subclinical psychotic experiences in healthy young adults: Associations with stress and genetic predisposition (2014) Genetic Testing and Molecular Biomarkers, 18 (10), pp. 683-689 53. Yu, Q., Zhang, R.-M., Jiang, Y., Wang, M., Yu, Y.-Q., Shi, J.-P. Meta-analysis on relationship between TAAR6 gene polymorphisms and schizophrenia (2014) Journal of Jilin University Medicine Edition, 40 (3), pp. 633-640 54. Ma, L., Wu, D.-D., Ma, S.L., Tan, L., Chen, X., Tang, N.L.S., Yao, Y.-G. Molecular evolution in the CREB1 signal pathway and a rare haplotype in CREB1 with genetic predisposition to schizophrenia (2014) Journal of Psychiatric Research, 57 (1), pp. 84-89 55. Su, L., Long, J., Liang, B., Gu, L., Pan, R., Li, L., Zhou, Y., Tang, X., Jiang, J., Chen, Q., Wei, B. Association between the NOTCH4 gene rs3131296 polymorphism with schizophrenia risk in the Chinese Zhuang population and Chinese Han population (2014) Acta Neuropsychiatrica, 26 (4), pp. 240-245 56. Zhang, S., Zhang, M., Zhang, J. An Exploratory Study on DRD2 and Creative Potential (2014) Creativity Research Journal, 26 (1), pp. 115-123 57. Shibuya, M., Watanabe, Y., Nunokawa, A., Egawa, J., Kaneko, N., Igeta, H., Someya, T. Interleukin 1 beta gene and risk of schizophrenia: Detailed case-control and family-based studies and an updated meta-analysis (2014) Human Psychopharmacology, 29 (1), pp. 31-37 58. Kapelski, P., Skibińska, M., Twarowska-Hauser, J. Association studies of interleukin's genes and their receptors in schizophrenia (2013) Psychiatria, 10 (1), pp. 9-18 59. Loh, H.C., Chow, T.J., Tang, P.Y., Yong, H.S. No association between AKT1 gene variants and schizophrenia: A Malaysian case-control study and meta-analysis (2013) Psychiatry Research, 209 (3), pp. 732-733 60. Ozomaro, U., Wahlestedt, C., Nemeroff, C.B. Personalized medicine in psychiatry: Problems and promises (2013) BMC Medicine, 11 (1), art. no. 132 61. Ohi, K., Hashimoto, R., Yasuda, Y., Fukumoto, M., Nemoto, K., Ohnishi, T., Yamamori, H., Takahashi, H., Iike, N., Kamino, K.,

Yoshida, T., Azechi, M., Ikezawa, K., Tanimukai, H., Tagami, S., Morihara, T., Okochi, M., Tanaka, T., Kudo, T., Iwase, M., Kazui, H., Takeda, M. The AKT1 gene is associated with attention and brain morphology in schizophrenia (2013) World Journal of Biological Psychiatry, 14 (2), pp. 100-113

62. Markett, S., Reuter, M., Montag, C., Weber, B. The dopamine D2 receptor gene DRD2 and the nicotinic acetylcholine receptor gene CHRNA4 interact on striatal gray matter volume: Evidence from a genetic imaging study (2013) NeuroImage, 64 (1), pp. 167-172
63. Fatjó-Vilas, M., Pomarol-Clotet, E., Salvador, R., Monté, G.C., Gomar, J.J., Sarró, S., Ortiz-Gil, J., Aguirre, C., Landín-Romero, R., Guerrero-Pedraza, A., Papiol, S., Blanch, J., McKenna, P.J., Fañanás, L. Effect of the interleukin-1β gene on dorsolateral prefrontal cortex function in schizophrenia: A genetic neuroimaging study (2012) Biological Psychiatry, 72 (9), pp. 758-765.
64. Yoshida, M., Shiroiwa, K., Mouri, K., Ishiguro, H., Supriyanto, I., Ratta-Apha, W., Eguchi, N., Okazaki, S., Sasada, T., Fukutake, M., Hashimoto, T., Inada, T., Arinami, T., Shirakawa, O., Hishimoto, A. Haplotypes in the expression quantitative trait locus of interleukin-1β gene are associated with schizophrenia (2012) Schizophrenia Research, 140 (1-3), pp. 185-191
65. Manceau, V., Kremmer, E., Nabel, E.G., Maucuer, A. The protein kinase KIS impacts gene expression during development and fear conditioning in adult mice (2012) PLoS ONE, 7 (8), art. no. e43946
66. Ram Murthy, A., Purushottam, M., Kiran Kumar, H.B.G., Vallikiran, M., Krishna, N., Jayramu Sriharsha, K., Janardhan Reddy, Y.C., Ghosh, S., Jain, S. Gender-specific association of TSNAX/DISC1 locus for schizophrenia and bipolar affective disorder in South Indian population (2012) Journal of Human Genetics, 57 (8), pp. 523-530.
67. Pishva, E., Kenis, G., Lesch, K.P., Prickaerts, J., Steinbusch, H.M.W., La Van Den Hove, D., Van Os, J., Rutten, B.P. Epigenetic epidemiology in psychiatry: A translational neuroscience perspective (2012) Translational Neuroscience, 3 (2), pp. 196-212
68. Mathieson, I., Munafò, M.R., Flint, J. Meta-analysis indicates that common variants at the DISC1 locus are not associated with schizophrenia (2012) Molecular Psychiatry, 17 (6), pp. 634-641
69. Girard, S.L., Dion, P.A., Rouleau, G.A. Schizophrenia genetics: Putting all the pieces together (2012) Current Neurology and Neuroscience Reports, 12 (3), pp. 261-266
70. Ohi, K., Hashimoto, R., Yasuda, Y., Fukumoto, M., Yamamori, H., Umeda-Yano, S., Okada, T., Kamino, K., Morihara, T., Iwase, M., Kazui, H., Numata, S., Ikeda, M., Ohnuma, T., Iwata, N., Ueno, S.-I., Ozaki, N., Ohmori, T., Arai, H., Takeda, M. Functional genetic variation at the NRGN gene and schizophrenia: Evidence from a gene-based case-control study and gene expression analysis (2012) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 159 B (4), pp. 405-413
71. Voisey, J., Swagell, C.D., Hughes, I.P., Lawford, B.R., Young, R.M., Morris, C.P. A novel DRD2 single-nucleotide polymorphism associated with schizophrenia predicts age of onset: HapMap Tag-single-nucleotide polymorphism analysis (2012) Genetic Testing and Molecular Biomarkers, 16 (2), pp. 77-81
72. Huertas, E., Bühler, K.-M., Echeverry-Alzate, V., Giménez, T., López-Moreno, J.A. C957T polymorphism of the dopamine D2 receptor gene is associated with motor learning and heart rate (2012) Genes, Brain and Behavior, 11 (6), pp. 677-683
73. Morris, C.P., Baune, B.T., Domschke, K., Arolt, V., Swagell, C.D., Hughes, I.P., Lawford, B.R., Young, R.M., Voisey, J. KPNA3 variation is associated with schizophrenia, major depression, opiate dependence and alcohol dependence (2012) Disease Markers, 33 (4), pp. 163-170.
74. Cho, A.R., Lee, S.M., Kang, W.S., Kim, S.K., Chung, J.-H. Assessment between Dopamine Receptor D2 (DRD2) polymorphisms and schizophrenia in Korean population (2012) Clinical Psychopharmacology and Neuroscience, 10 (2), pp. 88-93

		75. Dumaine, A., Maucuer, A., Barbet, A., Manceau, V., Deshommes, J., Méary, A., Szöke, A., Schürhoff, F., Llorca, P.-M., Lancon, C., Leboyer, M., Jamain, S. Genetic and molecular exploration of UHMK1 in schizophrenic patients (2011) Psychiatric Genetics, 21 (6), pp. 315-318 76. Peerbooms, O.L.J., van Os, J., Drukker, M., Kenis, G., Hoogveld, L., de Hert, M., Delespaul, P., van Winkel, R., Rutten, B.P.F. Meta-analysis of MTHFR gene variants in schizophrenia, bipolar disorder and unipolar depressive disorder: Evidence for a common genetic vulnerability? (2011) Brain, Behavior, and Immunity, 25 (8), pp. 1530-1543 77. Kim, S.-G., Song, J.Y., Joo, E.-J., Jeong, S.H., Kim, S.H., Lee, K.Y., Lee, N.Y., Ahn, Y.M., Kim, Y.S., Roh, M.-S. No association of functional polymorphisms in Methlylenetetrahydrofolate reductase and the risk and minor physical anomalies of schizophrenia in Korean population (2011) Journal of Korean Medical Science, 26 (10), pp. 1356-1363. 78. Moens, L.N., de Rijk, P., Reumers, J., van den Bossche, M.J.A., Glassee, W., de Zutter, S., Lenaerts, A.-S., Nordin, A., Nilsson, L.-G., Castello, I., Norrback, K.-F., Goossens, D., van Steen, K., Adolfsson, R., Del-Favero, J. Sequencing of DISC1 pathway genes reveals increased burden of rare missense variants in schizophrenia patients from a Northern Swedish population (2011) PLoS ONE, 6 (8), art. no. e23450 79. Sasayama, D., Hori, H., Teraishi, T., Hattori, K., Ota, M., Iijima, Y., Tatsumi, M., Higuchi, T., Amano, N., Kunugi, H. Possible association between Interleukin-1beta gene and schizophrenia in a Japanese population (2011) Behavioral and Brain Functions, 7, art. no. 35 80. Mouaffak, F., Kebir, O., Chayet, M., Tordjman, S., Vacheron, M.N., Millet, B., Jaafari, N., Bellon, A., Olié, J.P., Krebs, M.-O. Association of Disrupted in Schizophrenia 1 (DISC1) missense variants with ultra-resistant schizophrenia (2011) Pharmacogenomics Journal, 11 (4), pp. 267-273 81. Allen, E.A., Liu, J., Kiehl, K.A., Gelernter, J., Pearlson, G.D., Perrone-Bizzozero, N.I., Calhoun, V.D. Components of cross-frequency modulation in health and disease (2011) Frontiers in Systems Neuroscience, (JULY 2011), art. no. 59 82. Porcelli, S., Fabbri, C., Drago, A., Gibiino, S., de Ronchi, D., Serretti, A. Genetics and antidepressant: Where we are (2011) Clinical Neuropsychiatry, 8 (2), pp. 99-150 83. Saetre, P., Vares, M., Werge, T., Andreassen, O.A., Arinami, T., Ishiguro, H., Nanko, S., Tan, E.C., Han, D.H., Roffman, J.L., Muntjewerff, J.-W., Jagodzinski, P.P., Kempisty, B., Hauser, J., Vilella, E., Betcheva, E., Nakamura, Y., Regland, B., Agartz, I., Hall, H., Terenius, L., Jönsson, E.G. Methylenetetrahydrofolate reductase (MTHFR) C677T and A1298C polymorphisms and age of onset in schizophrenia: A combined analysis of independent samples (2011) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 156 (2), pp. 215-224 84. Girard, S.L., Xiong, L., Dion, P.A., Rouleau, G.A. Where are the missing pieces of the schizophrenia genetics puzzle? (2011) Current Opinion in Genetics and Development, 21 (3), pp. 310-316 85. Mi, H., Thomas, P.D., Ring, H.Z., Jiang, R., Sangkuhl, K., Klein, T.E., Altman, R.B. Pharm GKB summary: Dopamine receptor D2 (2011) Pharmacogenetics and Genomics, 21 (6), pp. 350-356 86. Tiwari, A.K., Zai, C.C., Müller, D.J., Kennedy, J.L. Genetics in schizophrenia: Where are we and what next? (2010) Dialogues in Clinical Neuroscience, 12 (3), pp. 289-303 87. Yoshimi, A., Aleksic, B., Kawamura, Y., Takahashi, N., Yamada, S., Usui, H., Saito, S., Ito, Y., Iwata, N., Inada, T., Noda, Y., Yamada, K., Ozaki, N. Gene-wide association study between the methylenetetrahydrofolate reductase gene (MTHFR) and schizophrenia in the Japanese population, with an updated meta-analysis on currently available data (2010) Schizophrenia Research, 124 (1-3), pp. 216-222 88. Voisey, J., Swagell, C.D., Hughes, I.P., Lawford, B.R., Young, R.M., Morris, C.P. Analysis of HapMap tag-SNPs in dysbindin (DTNBP1)

		reveals evidence of consistent association with schizophrenia (2010) European Psychiatry, 25 (6), pp. 314-319 89. Xu, M., He, L. Convergent Evidence Shows a Positive Association of Interleukin-1 Gene Complex Locus with Susceptibility to Schizophrenia in the Caucasian population (2010) Schizophrenia Research, 120 (1-3), pp. 131-142 90. Sipilä, T., Kananen, L., Greco, D., Donner, J., Silander, K., Terwilliger, J.D., Auvinen, P., Peltonen, L., Lönnqvist, J., Pirkola, S., Partonen, T., Hovatta, I. An Association Analysis of Circadian Genes in Anxiety Disorders (2010) Biological Psychiatry, 67 (12), pp. 1163-1170 91. Saadat, M. N-methyl-D-aspartate receptor NR1 subunit gene (GRIN1) G1001C polymorphism and susceptibility to schizophrenia: A meta-analysis (2010) EXCLI Journal, 9, pp. 11-16 92. Dubertret, C., Bardel, C., Ramoz, N., Martin, P.-M., Deybach, J.-C., Adès, J., Gorwood, P., Gouya, L. A genetic schizophrenia-susceptibility region located between the ANKK1 and DRD2 genes (2010) Progress in Neuro-Psychopharmacology and Biological Psychiatry, 34 (3), pp. 492-499 93. Simpson, E.H., Kellendonk, C., Kandel, E. A Possible Role for the Striatum in the Pathogenesis of the Cognitive Symptoms of Schizophrenia (2010) Neuron, 65 (5), pp. 585-596 94. Bertolino, A., Taurisano, P., Pisciotta, N.M., Blasi, G., Fazio, L., Romano, R., Gelao, B., Lo Bianco, L., Lozupone, M., Di Giorgio, A., Caforio, G., Sambataro, F., Niccoli-Asabella, A., Papp, A., Ursini, G., Sinibaldi, L., Popolizio, T., Sadee, W., Rubini, G. Genetically determined measures of striatal D2 signaling predict prefrontal activity during working memory performance (2010) PLoS ONE, 5 (2), art. no. e9348 95. Ohi, K., Hashimoto, R., Yasuda, Y., Yoshida, T., Takahashi, H., Iike, N., Iwase, M., Kamino, K., Ishii, R., Kazui, H., Fukumoto, M., Takamura, H., Yamamori, H., Azechi, M., Ikezawa, K., Tanimukai, H., Tagami, S., Morihara, T., Okochi, M., Yamada, K., Numata, S., Ikeda, M., Tanaka, T., Kudo, T., Ueno, S.-i., Yoshikawa, T., Ohmori, T., Iwata, N., Ozaki, N., Takeda, M. The chitinase 3-like 1 gene and schizophrenia: Evidence from a multi-center case-control study and meta-analysis (2010) Schizophrenia Research, 116 (2-3), pp. 126-132. 96. Watanabe, Y., Nunokawa, A., Kaneko, N., Someya, T. A case-control association analysis of CABIN1 with schizophrenia in a Japanese population (2010) Journal of Human Genetics, 55 (3), pp. 179-181. 97. Bristow, G.C., Lane, T.A., Walker, M., Chen, L., Sei, Y., Hyde, T.M., Kleinman, J.E., Harrison, P.J., Eastwood, S.L. Expression of Kinase Interacting with Stathmin (KIS, UHMK1) in human brain and lymphoblasts: effects of schizophrenia and genotype (2009) Brain Research, 1301, pp. 197-206 98. Kellendonk, C. Modeling excess striatal D2 receptors in mice (2009) Progress in Brain Research, 179 (C), pp. 59-65 99. Cuesta, M., Cermakian, N., Boivin, D.B. Circadian clock genes and psychiatric disorders (2006) The Genetic Basis of Sleep and Sleep Disorders, pp. 351-364.
10	**Genome-wide gene expression profiles of thyroid carcinoma: Identification of molecular targets for treatment of thyroid carcinoma**. Nikolova DN, Zembutsu H, Sechanov T, Vidinov K, Kee LS, Ivanova R, **Becheva E**, Kocova M, Toncheva D, Nakamura Y. Oncol Rep. 2008 Jul; 20(1):105-21 PMID: 18575725 **ДОКТОРСКА СТЕПЕН**	1. Hypomethylation-mediated overexpression of ITGA2 stimulates cell invasion and migration of thyroid carcinoma. Chen, H; Zhang, CY; (...); Chen, CY. Jul 2023 HISTOLOGY AND HISTOPATHOLOGY. 38 (7) , pp.787-796 2. Functional Assessment of Cancer-Linked Mutations in Sensitive Regions of Regulators of G Protein Signaling Predicted by Three-Dimensional Missense Tolerance Ratio AnalysisS Montañez-Miranda, C; Perszyk, RE; (...); Hepler, JR Jan 1 2023 MOLECULAR PHARMACOLOGY 103 (1) , pp.21-37 3. SYTL5 Promotes Papillary Thyroid Carcinoma Progression by Enhancing Activation of the NF-κB Signaling Pathway Huang, D; Zeng, Y; (...); Yu, ST Nov 14 2022 ENDOCRINOLOGY 164 (1) 4. Polyphenon E Effects on Gene Expression in PC-3 Prostate Cancer Cells Carastro, LM; Vallebuona, EJ; (...); Park, JY Nov 2022 INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES 23 (22) 5. Transcriptome Profile of Thyroid Glands in Bile Duct Ligation

		Mouse Model Jo, D; Kim, HK; (...); Song, J Aug 2022 INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES 23 (15) 6. MOLECULAR MARKERS PREDICTIVE OF LYMPH NODE INVOLVEMENT IN PAPILLARY THYROID CARCINOMAS de Oliveira, MLL; da Costa, JVA; (...); Canevari, RD 2022 REVISTA UNIVAP 28 (59) 7. Increased gene expression of TIMP1 and CHI3L1 in fine-needle aspiration biopsy samples from papillary thyroid cancer as compared to benign nodules Dimitrova, I; Shinkov, A; (...); Kovatcheva, R Sep 2021 DIAGNOSTIC CYTOPATHOLOGY 49 (9) , pp.1045-1051 8. Inhibition of lncRNA DCST1-AS1 suppresses proliferation, migration and invasion of cervical cancer cells by increasing miR-874-3p expression Liu, JL; Zhang, J; (...); Hu, XJ Jan 2021 JOURNAL OF GENE MEDICINE 23 (1) 9. Impact of the Tumor Microenvironment on the Gene Expression Profile in Papillary Thyroid Cancer Oczko-Wojciechowska, M; Pfeifer, A; (...); Krajewska, J May 2020 PATHOBIOLOGY 87 (2) , pp.143-154 10. The Impact of Transcription Factor Prospero Homeobox 1 on the Regulation of Thyroid Cancer Malignancy Rudzinska, M and Czarnocka, B May 2020 INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES 21 (9) 11. Identification of potential novel biomarkers to differentiate malignant thyroid nodules with cytological indeterminate Wu, DD; Hu, SD; (...); Liu, SB Mar 12 2020 BMC CANCER 20 (1) 12. Interplay of cysteine exposure and global protein dynamics in small-molecule recognition by a regulator of G-protein signaling protein Mohammadi, M; Mohammadiarani, H; (...); Vashisth, H Feb 2019 PROTEINS-STRUCTURE FUNCTION AND BIOINFORMATICS 87 (2) , pp.146-156 13. MicroRNA-486 inhibits cell proliferation, invasion and migration via down-regulating the TENM1 expressions and affecting ERK and Akt signaling pathways and epithelial-to-mesenchymal transition in papillary thyroid carcinoma Sun, YH; Liu, ZF; (...); Yu, B 2019 EUROPEAN REVIEW FOR MEDICAL AND PHARMACOLOGICAL SCIENCES 23 (19) , pp.8429-8439 14. RGS4 Regulates Proliferation And Apoptosis Of NSCLC Cells Via microRNA-16 And Brain-Derived Neurotrophic Factor He, Z; Yu, LH; (...); An, YH 2019 ONCOTARGETS AND THERAPY 12 , pp.8701-8714 15. HORMAD2 methylation-mediated epigenetic regulation of gene expression in thyroid cancer Lin, QY; Hou, S; (...); Lin, CH Oct 2018 JOURNAL OF CELLULAR AND MOLECULAR MEDICINE 22 (10) , pp.4640-4652 16. Angiogenesis and evading immune destruction are the main related transcriptomic characteristics to the invasive process of oral tongue cancer Pérez-Valencia, JA; Prosdocimi, F; (...); Rumjanek, FD Jan 31 2018 SCIENTIFIC REPORTS 17. Computational Method for Biomedical Binary Data Analysis: Development of a Thyroid Disease Index Using a Brute-Force Search with MLR Analysis Lee, JK; Han, WS; (...); Yoon, CN Dec 2017 BULLETIN OF THE KOREAN CHEMICAL SOCIETY 38 (12) , pp.1392-1397 18. Post-translational Regulation of Radioactive Iodine Therapy Response in Papillary Thyroid Carcinoma Amit, M; Na'ara, S; (...); Gil, Z Dec 2017 JNCI-JOURNAL OF THE NATIONAL CANCER INSTITUTE 109 (12) 19. Overexpression of teneurin transmembrane protein 1 is a potential marker of disease progression in papillary thyroid carcinoma Cheng, SP; Chen, MJ; (...); Liu, CL Nov 2017 CLINICAL AND EXPERIMENTAL MEDICINE 17 (4) , pp.555-564 20. Functional analysis of a novel, thyroglobulin-embedded microRNA gene deregulated in papillary thyroid carcinoma Kolanowska, M; Wójcicka, A; (...); Jazdzewski, K Aug 30 2017 SCIENTIFIC REPORTS 7 21. Long Noncoding RNA and mRNA Expression Profiles in the Thyroid Gland of Two Phenotypically Extreme Pig Breeds Using

		Ribo-Zero RNA Sequencing Shen, YF; Mao, HG; (...); Xu, NY Jul 2016 GENES 7 (7) 22. Gene expression profiling in undifferentiated thyroid carcinoma induced by high-dose radiation Bang, HS; Choi, MH; (...); Choi, SJ Jun 2016 JOURNAL OF RADIATION RESEARCH 57 (3) , pp.238-249 23. A three-gene panel that distinguishes benign from malignant thyroid nodules Zheng, B; Liu, J; (...); Lu, H Apr 1 2015 INTERNATIONAL JOURNAL OF CANCER 136 (7) , pp.1646-1654 24. Connecting gene expression data from connectivity map and in silico target predictions for small molecule mechanism-of-action analysis Ravindranath, AC; Perualila-Tan, N; (...); Bender, A Jan 2015 MOLECULAR BIOSYSTEMS 11 (1) , pp.86-96 25. Biomarkers in Diagnosis of Papillary Thyroid Carcinoma Cañadas-Garre, M; Pérez, NM; (...); Llamas-Elvira, JM 2014 CANCER BIOMARKERS: MINIMAL AND NONINVASIVE EARLY DIAGNOSIS AND PROGNOSIS , pp.679-750 26. Identification of Novel Gene Targets and Functions of p21-Activated Kinase 1 during DNA Damage by Gene Expression Profiling Motwani, M; Li, DQ; (...); Kumar, R Aug 12 2013 PLOS ONE 8 (8) 27. Thyroid Regeneration: Characterization of Clear Cells After Partial Thyroidectomy Ozaki, T; Matsubara, T; (...); Kimura, S May 2012 ENDOCRINOLOGY 153 (5) , pp.2514-2525 28. Gene expression profiling of papillary thyroid carcinomas in Korean patients by oligonucleotide microarrays Chung, KW; Kim, SW and Kim, SW May 2012 JOURNAL OF THE KOREAN SURGICAL SOCIETY 82 (5) , pp.271-280 29. Three-Gene Molecular Diagnostic Model for Thyroid Cancer Prasad, NB; Kowalski, J; (...); Zeiger, MA Mar 2012 THYROID 22 (3) , pp.275-284 30. Genetic Variations in the Regulator of G-Protein Signaling Genes Are Associated with Survival in Late-Stage Non-Small Cell Lung Cancer Dai, JY; Gu, J; (...); Wu, XF Jun 17 2011 PLOS ONE 6 (6) 31. Predicting response of bladder cancers to gemcitabine and carboplatin neoadjuvant chemotherapy through genome-wide gene expression profiling Kato, Y; Zembutsu, H; (...); Nakamura, Y Jan-feb 2011 EXPERIMENTAL AND THERAPEUTIC MEDICINE 2 (1) , pp.47-56 32. RNA-binding protein HuR regulates RGS4 mRNA stability in rabbit colonic smooth muscle cells Li F, Hu DY, Liu S, Mahavadi S, Yen W, Murthy KS, Khalili K, Hu W. RNA-binding protein HuR regulates RGS4 mRNA stability in rabbit colonic smooth muscle cells. Am J Physiol Cell Physiol 299: C1418-C1429, 2010. First published September 29, 2010; doi:10.1152/ajpcell.00093.2010.- 33. Genetic variations in regulator of G-protein signaling genes as susceptibility loci for second primary tumor/recurrence in head and neck squamous cell carcinoma Wang, JM; Lippman, SM; (...); Wu, XF Oct 2010 CARCINOGENESIS 31 (10) , pp.1755-1761 34. Cloning and characterization of rabbit Rgs4 promoter in gut smooth muscle Li, F; Murthy, KS; (...); Hu, WH Feb 1 2010 GENE 451 (1-2) , pp.45-53 35. A multigene predictor of metastatic outcome in early stage hormone receptor-negative and triple-negative breast cancer Yau, C; Esserman, L; (...); Benz, CC 2010 BREAST CANCER RESEARCH 12 (5) 36. Potential Utility and Limitations of Thyroid Cancer Cell Lines as Models for Studying Thyroid Cancer Pilli, T; Prasad, KV; (...); Prabhakar, BS Dec 2009 THYROID 19 (12) , pp.1333-1342 37. Regulator of G-protein signaling (RGS) proteins in cancer biology Hurst, JH and Hooks, SB Nov 15 2009 BIOCHEMICAL PHARMACOLOGY 78 (10) , pp.1289-1297 38. Cellular distributions of molecules with altered expression specific to thyroid proliferative lesions developing in a rat thyroid carcinogenesis model Woo, GH; Takahashi, M; (...); Shibutani, M Apr 2009 CANCER SCIENCE 100 (4) , pp.617-625

СПИСЪК

на цитирания на научни публикации на д-р Елица Бечева - Крайчир

ORCiD 0009-0009-8339-5637 / Web of Science Researcher ID: KLC-9167-2024 / Scopus ID 56662187800

СПИСЪК

на цитирания в нереферирани публикации
на д-р Елица Бечева - Крайчир

№	Цитирана публикация	Цитираща публикация
1	**Skewed X-inactivation in a family with *DLG3*-associated X-linked intellectual disability**. Gieldon L., Mackenroth L., **Betcheva-Krajcir E**., Rump A., Beck-Wodl S., Schallner J., Di Donato N., Schröck, E., Tzschach A. (**2017**) American Journal of Medical Genetics, Part A, 173 (9) , pp. 2545-2550.	1. Meryem Alagoz, Nasim Kherad, Huseyin Avni Solgun et al. DLG3 Impairment Caused by Missense Variants in Non-Syndromic X-Linked Mental Retardation, 16 July 2021, PREPRINT (Version 1) available at Research Square [https://doi.org/10.21203/rs.3.rs-692598/v1] 2. Alotaibi M (2021) De Novo Mutations in DLG3 in Girl with Intellectual Disabilities in Saudi. Open J Case Rep 2: 129
3	**Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH**. Hackmann K., Kuhlee F., **Betcheva-Krajcir E**., Kahlert A.-K., Mackenroth L., Klink B., Di Donato N., Tzschach, A., Kast, K., Wimberger, P., Schrock, E., Rump A. (**2016**) Breast Cancer Research and Treatment, 159 (3) , pp. 585-590.	1. Lantieri, F.; Malacarne, M.; Gimelli, S.; Santamaria, G.; Coviello, D.; Ceccherini, I. Custom Array Comparative Genomic Hybridization: the Importance of DNA Quality, an Expert Eye, and Variant Validation. *Int. J. Mol. Sci.* 2017, *18*, 609. https://doi.org/10.3390/ijms18030609 2. Ernst, Corinna. Computational methods for improved cancer risk prediction based on multi-gene panel analysis in a routine diagnostic setting. PhD thesis, Universität zu Köln. 2021. ORCID: 0000-0001-7756-8815. https://kups.ub.uni-koeln.de/52448/ 3. Alothman, Sahar J. Studies of Breast Cancer Chemoprevention in Human High-risk Primary Cells and Genetically Engineered Mouse Models. A Dissertation submitted to the Faculty of the Graduate School of Arts and Science Georgetown University ProQuest Dissertations Publishing, 2018. 10793920. 4. Pelttari, Liisa. Genetics of Breast and Ovarian Cancer Predisposition with a Focus on RAD51C and RAD51D Genes. 2018-08-14. http://urn.fi/URN:ISBN:978-951-51-4374-7. ISBN 978-951-51-4374-7
4	**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 J.D., Janavicius R., Machackova E., Foretova 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. (**2016**) PLoS Genetics, 12 (8) , art. no. e1006248	1. Chevalier, Aaron. Methods of Mutational Signature Analysis for Discovery, Comparison, and Drug Response Prediction. Dissertation Boston University ProQuest Dissertations Publishing, 2022.29256333. 2. Král, Jan. Identifikace dědičných faktorů ovlivňujících vznik karcinomu pankreatu a dalších solidních tumorů. Dizertační práce, vedoucí Janatová, Markéta. Praha: Univerzita Karlova, 1. lékařská fakulta, Ústav biochemie a experimentální onkologie 1. LF UK, 2023. 3. Koudová M., Puchmajerová A. Rizika solidních nádorů u heterozygotních přenašečů recesivních syndromů. Risks of Solid Tumors in Heterozygous Carriers of Recessive Syndromes. Klin Onkol 2019; 32 (Suppl 2): 2S14–2S 23doi: 10.14735/amko2019S14. 4. Cooney,Kathleen A.; Tomlins,Scott. High-Throughput Sequencing of Germline and Tumor From Men with Early-Onset Metastatic Prostate Cancer. Technical Report,30 Sep 2013,29 Sep 2016. Regents of the University of Michigan Ann Arbor United States. U.S. Army Medical Research and Materiel Command Fort Detrick, Maryland 21702-5012 5. Chmelařová, Žaneta. Analýza genetických faktorů vzniku karcinomu prsu. Diplomová práce, vedoucí Kleiblová, Petra. Univerzita Karlova, Přírodovědecká fakulta, Katedra antropologie a genetiky člověka, 2018. 6. Jean-Christophe Thery. Détection et contribution de variants rares constitutionnels dans les formes précoces de cancer du sein : Apports du Séquençage de Nouvelle Génération.. Médecine humaine et pathologie. Normandie Université, 2019. Français. ⟨NNT : 2019NORMR111⟩. ⟨tel-02501260⟩
5	**HBOC multi-gene panel testing: comparison of two sequencing centers**. Schroeder C., Faust U., Sturm M., Hackmann K., Grundmann K., Harmuth F., Bosse K., Kehrer, M., Benkert, T., Klink, B., Mackenroth, L., **Betcheva-Krajcir, E**., Wimberger, P., Kast, K., Heilig, M., Nguyen, H., Riess, O., Schröck, E., Bauer,	1. Trivedi MS, Crew KD. Implications of multigene testing for hereditary breast cancer in primary care. World J Obstet Gynecol 2016; 5(1): 50-57 [DOI: 10.5317/wjog.v5.i1.50] 2. Alcántara Gutiérrez, Alejandro; Cárceles Guillén, Pedro José; Grado. En MedicinaIncidencia del síndrome de cáncer de mama y ovario hereditario en el área de salud del Hospital Universitario Nuestra Señora de Candelaria. 2022. Repositorio institucional de la

	P., Rump, A. (**2015**) Breast Cancer Research and Treatment, 152 (1) , pp. 129-136.	Universidad de La Laguna. info:eu-repo/semantics/bachelorThesis. http://riull.ull.es/xmlui/handle/915/27034 3. Helms, Malina. Die Tumorgenetische Spezialsprechstunde im Zentrum Familiärer Brust- und Eierstockkrebs der Charité. Charakteristika der Ratsuchenden in den Jahren 2016 und 2017. 2021. Dissertation. 610 Medizin und Gesundheit. Charité - Universitätsmedizin Berlin. https://refubium.fu-berlin.de/handle/fub188/30731. http://dx.doi.org/10.17169/refubium-30470. urn:nbn:de:kobv:188-refubium-30731-4 4. da Costa E Silva Carvalho S, Cury NM, Brotto DB, de Araujo LF, Rosa RCA, Texeira LA, Plaça JR, Marques AA, Peronni KC, Ruy PC, Molfetta GA, Moriguti JC, Carraro DM, Palmero EI, Ashton-Prolla P, de Faria Ferraz VE, Silva WA Jr. Germline variants in DNA repair genes associated with hereditary breast and ovarian cancer syndrome: analysis of a 21 gene panel in the Brazilian population. BMC Med Genomics. 2020 Feb 10;13(1):21. doi: 10.1186/s12920-019-0652-y. PMID: 32039725; PMCID: PMC7011249. 5. Tuncer, S. B., & ÇELİK, B., (2021). Clinical and Molecular Characteristics and Genetic Risks of Ovarian Cancer. Academic Developments on Health Sciences (pp.327-345), İzmir: Duvar Yayınları. 6. Pierre Fermey. Identification de nouvelles bases moléculaires des cancers précoces par séquençage à haut débit.. Génétique humaine. Normandie Université, 2017. Français. ⟨NNT : 2017NORMR110⟩. ⟨tel-01744221⟩ 7. Kraemer D, Azzarello-Burri S, Steindl K, Boonsawat P, Zweier M, Dedes KJ, Joset P, Fink D, Rauch A. Prevalence of genetic susceptibility for breast and ovarian cancer in a non-cancer related study population: secondary germline findings from a Swiss single centre cohort. Swiss Med Wkly [Internet]. 2019 Aug. 18 [cited 2024 May 16];149(3334):w20092. Available from: https://smw.ch/index.php/smw/article/view/2658 8. Kulkarni SS, Nag S, Patra A, Pant HB, Agiwal V, Nirupama AY, et al. Prevalence of germline mutations in women with Breast and/or Ovarian cancer in a tertiary care center in Pune, India. Int J Mol Immuno Oncol 2023;8:65-71. 9. Özge Şebnem Çıldır, Özge Özmen, Selim Kul, Ali Rişvanlı, Gözde Özalp, Ahmet Sabuncu, Oğuz Kul. Genetic analysis of PALB2 gene WD40 domain in canine mammary tumour patients. 25 March 2024. https://doi.org/10.1002/vms3.1366. Veterinary Medicine and Science published by John Wiley & Sons Ltd. Volume10, Issue3 May 2024 e1366 10. Oda, K., Tanikawa, M., Sone, K. et al. Recent advances in targeting DNA repair pathways for the treatment of ovarian cancer and their clinical relevance. Int J Clin Oncol 22, 611–618 (2017). https://doi.org/10.1007/s10147-017-1137-7 11. Castillo Guardiola, Verónica. Aportación de la secuenciación masiva de nueva generación en el diagnóstico del síndrome de cáncer de mama y ovario hereditario mediante el uso de un panel de genes.12-jul-2019 http://hdl.handle.net/10201/73883. Ciencias de la Salud. Escuela Internacional de Doctorado 12. Mestre Terkemani, Younes. Utilidad del estudio genético, mediante un amplio panel de genes clínicamente accionables, en el síndrome de cáncer de mama y ovario hereditario. 15-nov-2023 http://hdl.handle.net/10201/135777. Ciencias de la Salud. Escuela Internacional de Doctorado 13. Dixon, K. (2020). Molecular characterization and genetic diagnosis of cancer predisposition syndromes using genome and transcriptome sequencing (T). University of British Columbia. Retrieved from https://open.library.ubc.ca/collections/ubctheses/24/items/1.0394119 14. Zilski, Nicole. Analyse der Lebensqualität mit und ohne prophylaktischer Ovarektomie bei BRCA1/2-Mutationsträgerinnen. 2023 610 Medizin und Gesundheit. Dissertation. Charité - Universitätsmedizin Berlin. https://refubium.fu-berlin.de/handle/fub188/37418. http://dx.doi.org/10.17169/refubium-37131. urn:nbn:de:kobv:188-refubium-37418-2 15. Bárbara Alemar Beserra Temes. Síndromes de predisposição hereditária ao câncer de mama e/ou ovário: análises genômicas,

		epidemiologia molecular e caracterização clínica. Thesis. Porto Alegre, dezembro de 2017. UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL 16. Mateja Smogavec. Identifizierung von Patientinnen und Patienten mit der hereditären Form des Mamma- und Ovarialkarzinoms mittels Next-Generation-Sequencing-(NGS)-Technologie. 2018. Institut für Humangenetik (Prof. Dr. med. B. Wollnik) der Medizinischen Fakultät der Universität Göttingen. DISSERTATION. Georg-August-Universität zu Göttingen
6	**Whole-genome-wide association study in the Bulgarian population reveals *HHAT* as schizophrenia susceptibility gene**. **Betcheva E.T.**, Yosifova A.G., Mushiroda T., Kubo M., Takahashi A., Karachanak S.K., Zaharieva I.T., Hadjidekova, S., Dimova, I., Vazharova, R., Stoyanov, D., Nakamura Y. (**2013**) Psychiatric Genetics, 23 (1), pp. 11-19.	1. Hawi Z, Tong J, Dark C, Yates H, Johnson B, Bellgrove MA. The role of cadherin genes in five major psychiatric disorders: A literature update. Am J Med Genet Part B. 2018; 177B: 168–180. https://doi.org/10.1002/ajmg.b.32592 2. Pol-Fuster Josep , Cañellas Francesca , Ruiz-Guerra Laura , Medina-Dols Aina , Bisbal-Carrió Bàrbara , Asensio Víctor , Ortega-Vila Bernat , Marzese Diego , Vidal Carme , Santos Carmen , Lladó Jerònia , Olmos Gabriel , Heine-Suñer Damià , Strauch Konstantin , Flaquer Antònia , Vives-Bauzà Cristòfol. Familial Psychosis Associated With a Missense Mutation at MACF1 Gene Combined With the Rare Duplications DUP3p26.3 and DUP16q23.3, Affecting the CNTN6 and CDH13 Genes. Frontiers in Genetics. VOLUME 12. 2021.https://www.frontiersin.org/journals/genetics/articles/10.3389/fgene.2021.622886. DOI 10.3389/fgene.2021.622886. ISSN 1664-8021 3. Josep Pol Fuster. Disentangling the genomic architecture of psychosis in families with high prevalence through system genomic approaches. Thesis. Programa de Doctorado en Biotecnología Biomédica y Evolutiva por la Universidad de las Illes Balears. 2021. https://dialnet.unirioja.es/servlet/tesis?codigo=294147 4. Alexandros Rammos.Interpretation and improvement of the current genetic epidemiology methodology for schizophrenia. A thesis submitted for the degree of Doctor of Philosophy (PhD) to the School of Genetics and Microbiology, Department of Genetics, Trinity College Dublin December 2017
7	**Methylenetetrahydrofolate reductase (*MTHFR*) C677T and A1298C polymorphisms and age of onset in schizophrenia: A combined analysis of independent samples**. Saetre P., Vares M., Werge T., Andreassen O.A., Arinami T., Ishiguro H., Nanko S., Tan, Ch., Han, D., Roffman, J., Muntjewerff, Jan-Willem, Jagodzinski, P., Kempisty, B., Hauser, J., Vilella, **E., Betcheva**, E., Nakamura, Y., Regland, B., Agartz, I., Hall, H., Terenius, L., Jonsson E.G. (**2011**) American Journal of Medical Genetics, Part B: Neuropsychiatric Genetics, 156 (2), pp. 215-224.	1. Evinova A, Babusikova E, Straka S, Ondrejka I, Lehotsky J. Analysis of genetic polymorphisms of brain-derived neurotrophic factor and methylenetetrahydrofolate reductase in depressed patients in a Slovak (Caucasian) population. Gen Physiol Biophys. 2012 Dec;31(4):415-22. doi: 10.4149/gpb_2012_049. PMID: 23255668. 2. Audiffred Jaramillo, Ricardo Ignacio, García Cobián, Teresa Arcelia, García de Alba García, Javier Eduardo, & Rubio Arellano, Edy David. (2016). Asociación del polimorfismo MTHFR C677T con la predominancia sintomatológica en una muestra de pacientes con esquizofrenia de Jalisco, México. Acta universitaria, 26(1), 56-64. https://doi.org/10.15174/au.2016.838 3. Audiffred Jaramillo, Ricardo Ignacio, García Cobián, Teresa Arcelia, García de Alba García, Javier Eduardo, & Rubio Arellano, Edy David. (2016). Asociación del polimorfismo MTHFR C677T con la predominancia sintomatológica en una muestra de pacientes con esquizofrenia de Jalisco, México. Acta universitaria, 26(1), 56-64. https://doi.org/10.15174/au.2016.838 4. Mărginean, Corina; Procopciuc, Lucia; Fodoreanu, Liana. Implicațiile polimorfismelor genei MTHFR în etiologia schizofreniei.Psihiatru.ro, 2017, Vol 49, Issue 2, p45. ISSN 1841-4877. Academic Journal 10.26416/Psih.49.2.2017.1107. EBSCO 5. Ricardo Ignacio Audiffred Jaramillo, Teresa Arcelia García Cobián, Javier Eduardo García de Alba García, Edy David Rubio Arellano. Asociación del polimorfismo MTHFR C677T con la predominancia sintomatológica en una muestra de pacientes con esquizofrenia de Jalisco, México. Acta Universitaria. Volumen 26 No. 1 doi: 10.15174/au.2016.838 I S S N (online) 2 0 0 7 - 9 6 2 1 0 6. Tan, Jaclyn Ai Chin and Geetha Gunasekaran, and Mohd Hanafi Ahmad Damanhuri, and Chan, Lai Fong and Loo, Jiann Lin and Goon, Jo Aan (2019) Oxidative status in bipolar disorder (BD) and its correlation with age, gender and body mass index (BMI). Sains Malaysiana, 48 (5). pp. 1083-1095. ISSN 0126-6039