

PUBLIKACJE:

Charzewska A, Terczyńska I, Lipiec A, Mazurczak T, Górka-Skoczylas P, Szlendak R, Kanabus K, Tataj R, Dawidziuk M, Wojtaś B, Gielniewski B, Bal J, Stawicka E, Hoffman-Zacharska D (2023) **Genetic risk factors for neurological disorders in children with adverse events following immunization: a descriptive study of Polish case series.** *Int J Mol Sci* 24(2):1117. doi: 10.3390/ijms24021117. (IF=6.208) (MNiSW= 140)

Zayat V, Kuczynska Z, Liput M, Metin E, Rzońca-Niewczas S, Smyk M, Mazurczak T, Goszczanska-Ciuchta A, Hoffman-Zacharska D, Buzanska L (2023) **Generation of ventral forebrain organoids from human induced pluripotent stem cell lines derived from patients with distinct SCN1A mutations.** *Cells* 12(2):339. doi: 10.3390/cells12020339. (IF=7.66) (MNiSW= 140)

Leonardi E, Aspromonte MC, Drongitis D, Bettella E, Verrillo L, Polli R, McEntagart M, Licchetta L, Dilena R, D'Arrigo S, Ciaccio C, Esposito S, Leuzzi V, Torella A, Baldo D, Lonardo F, Bonato G, Pellegrin S, Stanzial F, Posmyk R, Kaczorowska E, Carecchio M, Gos M, Rzońca-Niewczas S, Miano MG, Murgia A (2023) **Expanding the genetics and phenotypic spectrum of Lysine-specific demethylase 5C (KDM5C): a report of 13 novel variants.** *Eur J Hum Genet.* 31(2):202-215. (IF=5,351) (MNiSW=100)

Calame DG; Guo T, Wang Ch, Garrett L, Jolly A, Dawood M, Kurolap A, Henig NZ, Fatih JM; Herman I; Du H; Mitani T; Becker L, Rathkolb B, Seisenberger C, Marschall S, Hunter JV; Gerard A, Heidlebaugh A; Challman T, Spillmann R; Jhangiani SN, Coban-Akdemir Z, Lalani S, Revah-Politi A, Iglesias A, Guzman E, Baugh E, Boddaert N; Rondeau S; Clothide O; Barcia G; Tan QKG, Thiffault I, Sheikh K, Biliciler S, Mei D, Melani F, Shashi V, Yaron Y, Steele M, Wakeling E, Østergaard E, Nazaryan-Petersen L, Undiagnosed Diseases Network, Torti E, Thevenon J, Bruel A-L, Thauvin-Robinet C, Popp D, Platzer K, Mokry J, Gawlinski P, Wiszniewski W, Marafi D; Pehlivan D; Posey JE; Gibbs RA; Gailus-Durner V; Guerrini R, Fuchs H, de Angelis MH, Hölter SM, Cheung H-H, Gu S, Lupski JR (2023) **Monoallelic variation in DHX9, the gene encoding the DEXH-box helicase DHX9, underlies neurodevelopmental disorders and Charcot-Marie-Tooth disease.** *Am J Hum Genet* S0002-110(8):1394-1413 (IF=11,043) (MNiSW=200)

Werren EA, Guxholli A, Orenstein N, Narayanan V, Rafiullah R, Dawidziuk M, Wiszniewski W, Gawlinski P, Umair M, Khan A, Genevieve D, Lehalle D, van Gassen KLI, Giltay JC, Rappold GA, University of Washington Center for Mendelian Genomics (Keegan CE, Bielas SL, Srivastava A (2023). **Biallelic variants in CSMD1 cause neurodevelopmental defects and intellectual disability.** *Genetics in Medicine.* (IF=8,822) (MNiSW=200)

Niepokój K, Rygiel AM, Wertheim-Tysarowska K, Sawicka J, Obersztyn E, Kutkowska-Kaźmierczak A, Klapecki J, Barczyk A, Własienko P, Jurczak P, Lebedzińska A, Śmigiel R, Jakubiak A, Wierzba J, Kaczorowska E, Pietrzyk A, Sorbaj-Sucharska G, Limon J, Bal J (2023) **NGS sequencing reveals the cause of hearing loss in a group of Polish patients with an isolated non-DFNB1 hearing loss.** *Genes* (IF=4,141) (MNiSW=100)

Wertheim-Tysarowska K, Osipowicz K, Gielniewski B, Wojtaś B, Szabelska-Beręsewicz A, Zyprych-Walczak J, Mika A, Tysarowski A, Duk K, Rygiel AM, Niepokój K, Woźniak K, Kowalewski C, Wierzba J, Jezela-Stanek A (2023) **The epidermal transcriptome analysis of a novel c.639_642dup LORICRIN variant - delineation of the lorocrin keratoderma pathology.** *Int J Mol Sci* 9;24(11):9459. doi: 10.3390/ijms24119459 (IF=6,208) (MNiSW=140)

Himmelreich N, Bertoldi M, Alfadhel M, Alghamdi MA, Anikster Y, Bao X, Bashiri FA, Zeev BB, Bisello G, Ceylan AC, Chien YH, Choy YS, Elsea SH, Flint L, García-Cazorla À, Gijavanekar C, Gümüş EY, Hamad MH, Hişmi B, Honzik T, Hübschmann OK, Hwu WL, Ibáñez-Micó S, Jeltsch K, Juliá-Palacios N, Kasapkara ÇS, Kurian MA, Kusmierska K, Liu N, Ngu LH, Odom JD, Ong WP, Opladen T, Oppeboen M, Pearl PL, Pérez

B, Pons R, Rygiel AM, Shien TE, Spaul R, Sykut-Cegielska J, Tabarki B, Tangeraas T, Thöny B, Wassenberg T, Wen Y, Yakob Y, Yin JGC, Zeman J, Blau N (2023) **Prevalence of DDC genotypes in patients with aromatic L-amino acid decarboxylase (AADC) deficiency and in silico prediction of structural protein changes.** Mol Genet Metab. 2;139(3):107624. doi: 10.1016/j.ymgme.2023.107624. (IF=4,204) (MNiSW=100)

Jaczyńska R, Bekiesinska-Figatowska M, Sobieraj P, Issat T, Gos M, Obersztyn E (2023) **Prenatal and neonatal ultrasound and magnetic resonance imaging diagnosis of Sprengel's deformity with unusual associations.** Fetal Diagn Ther. doi: 10.1159/000531677 (IF=2,208) (MSWiA=100)

Pankiewicz K, Chotkowska E, blagoska B, Gos M, Issat T (2023) **COVID-19-related premature ovarian insufficiency: case report and literature review.** Climacteric doi.org/10.1080/13697137.2023.2246878 (IF=2,8) (MNiSW=70)

Dawidziuk M, Podwysocka A, Jurek M, Obersztyn E, Bekesińska-Figatowska M, Goszczynska-Ciuchta A, Bukowska-Olech E, Rygiel AM, Guilbridge DL, Wiszniewski W, Gawlinski P (2022) **Congenital coenzyme Q5-linked pathology: causal genetic association cre phenotype, and molecular mechanism.** JAG 64(3):507-514 (IF= 2,4) (MNiSW= 140)

Jankowska KK, Kutkowska-Kazmierczak A, Rygiel A (2023) **Hypogonadism - when does genetic diagnosis help in therapy?** Ginekol Pol. doi: 10.5603/gpl.97327. (IF=1.3) (MNiSW=70)

Bonardi CM, Møller RS, Ruiz-Reig N, Chai G, Madsen CG, Bayat A, Hammer TB, Fenger CD, Gardella E, Gawlinski P, Dawidziuk M, Wiszniewski W, Rossi M, Lesca G, Gouy E, Jepsen B, Mieszczanek TS, Aittaleb M, Brusgaard K, Tissir F, Rubboli G (2023) **Biallelic variants in CELSR1 cause brain malformations, neurodevelopmental delay, and epilepsy in humans.** Journal of Clinical Investigation (IF = 19.477) (MNiSW= 200)

Bielas S, Werren E, Guxholli A, Orenstein N, Narayanan, Rafiullah R, Dawidziuk M, Wiszniewski W, Gawlinski P, Umair M, Khan A, Genevieve D, Lehalle D, Giltay J, van Gassen KLI, Rappold G, Keegan C, Srivastava A (2023) **Biallelic variants in CSMD1 are implicated in a neurodevelopmental disorder with intellectual disability and variable cortical malformations.** Cell Death & Disease (IF = 9,696) (MNiSW=140)

Freud LR, Galloway S, Crowley TB, Moldenhauer J, Swillen A, Breckpot J, Borrell A, Vora NL, Cuneo B, Hoffman H, Gilbert L, Nowakowska B, Geremek M, Kutkowska-Każmierczak A, Vermeesch JR, Devriendt K, Busa T, Sigaudy S, Vigneswaran T, Simpson JM, Dungan J, Gotteiner N, Gloning KP, Digilio MC, Unolt M, Putotto C, Marino B, Repetto G, Fadic M, Garcia-Minaur S, Achón Buil A, Thomas MA, Fruitman D, Beecroft T, Hui PW, Oskarsdóttir S, Bradshaw R, Criebaum A, Norton ME, Lee T, Geiger M, Dunnington L, Isaac J, Wilkins-Haug L, Hunter L, Izzi C, Toscano M, Ghi T, McGlynn J, Romana Grati F, Emanuel BS, Gaiser K, Gaynor JW, Goldmuntz E, McGinn DE, Schindewolf E, Tran O, Zackai EH, Yan Q, Bassett AS, Wapner R, McDonald-McGinn DM (2023). **Prenatal vs postnatal diagnosis of 22q11.2 deletion syndrome: cardiac and noncardiac outcomes through 1 year of age.** Am J Obstet Gynecol. S0002-9378(23)00611-7. doi: 10.1016/j.ajog.2023.09.005 (IF=10,693) (MNiSW= 140)

Smyk M, Geremek M, Ziemkiewicz K, Gambin T, Kutkowska-Każmierczak A, Kowalczyk K, Plaskota I, Wiśniowiecka-Kowalnik B, Bartnik-Głaska M, Niemiec M, Grad D, Piotrowicz M, Gieruszczak-Biatek D, Pietrzyk A, Crowley TB, Giunta V, McGinn DE, Zackai EH, Tran O, Emanuel BS, McDonald-McGinn DM, Nowakowska BA (2023) **Coexisting Conditions Modifying Phenotypes of Patients with 22q11.2 Deletion Syndrome.** Genes (Basel). 14(3):680. doi: 10.3390/genes14030680. PMID: 36980952 (IF=4,141) (MNiSW= 100)

Óskarsdóttir S, Boot E, Crowley TB, Loo JCY, Arganbright JM, Armando M, Baylis AL, Breetvelt EJ, Castelein RM, Chadehumbe M, Cielo CM, de Reuver S, Eliez S, Fiksinski AM, Forbes BJ, Gallagher E, Hopkins SE, Jackson OA, Levitz-Katz L, Klingberg G, Lambert MP, Marino B, Mascarenhas MR, Moldenhauer J, Moss EM, Nowakowska BA, Orchanian-Cheff A, Putotto C, Repetto GM, Schindewolf E, Schneider M, Solot CB, Sullivan KE, Swillen A, Unolt M, Van Batavia JP, Vingerhoets C, Vorstman J, Bassett AS, McDonald-McGinn

DM (2023) **Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome.** Genet Med. 25(3):100338. doi: 0.1016/j.gim.2022.11.006. PMID: 36729053 (IF=8,8) (MNiSW= 200)

Boot E, Óskarsdóttir S, Loo JCY, Crowley TB, Orchanian-Cheff A, Andrade DM, Arganbright JM, Castelein RM, Cserti-Gazdewich C, de Reuver S, Fiksinski AM, Klingberg G, Lang AE, Mascarenhas MR, Moss EM, Nowakowska BA, Oechslein E, Palmer L, Repetto GM, Reyes NGD, Schneider M, Silversides C, Sullivan KE, Swillen A, van Amelsvoort TAMJ, Van Batavia JP, Vingerhoets C, McDonald-McGinn DM, Bassett AS (2023) **Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome.** Genet Med. 25(3):100344. doi: 10.1016/j.gim.2022.11.012. PMID: 36729052 (IF=8,8) (MNiSW= 200)

Dobrzeńiecka W, Daca M, Nowakowska B, Sobiesiak M, Szewczyk-Golec K, Woźniak A, Hotyńska-Iwan I (2023) **The Impact of Diclofenac Gel on Ion Transport in the Rabbit (*Oryctolagus cuniculus*) Skin: An In Vitro Study.** Molecules. 28(3):1332. doi: 10.3390/molecules28031332. PMID: 36770998 (IF=4,927) (MNiSW= 140)

Blagowidow N, Nowakowska B, Schindewolf E, Grati FR, Putotto C, Breckpot J, Swillen A, Crowley TB, Loo JCY, Lairson LA, Óskarsdóttir S, Boot E, Garcia-Minaur S, Cristina Digilio M, Marino B, Coleman B, Moldenhauer JS, Bassett AS, McDonald-McGinn DM (2023) **Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions.** Genes (Basel). 14(1):160. doi: 10.3390/genes14010160. PMID: 36672900 (IF=4,141) (MNiSW= 100)

Bijok J, Dąbkowska S, Kucińska-Chahwan A, Massalska D, Nowakowska B, Gawlik-Zawiślak S, Panek G, Roszkowski T (2023) **Prenatal diagnosis of acrania/exencephaly/anencephaly sequence (AEAS): additional structural and genetic anomalies.** Arch Gynecol Obstet. 307(1):293-299. doi: 10.1007/s00404-022-06584-3. PMID: 35554661 (IF=2,493) (MNiSW= 70)

Bzdęga K, Kutkowska-Kaźmierczak A, Deutsch GH, Plaskota I, Smyk M, Niemiec M, Barczyk A, Obersztyn E, Modzelewski J, Lipska I, Stankiewicz P, Gajecka M, Rydzanicz M, Płoski R, Szczapa T, Karolak JA (2023) **Prenatal Detection of a FOXF1 Deletion in a Fetus with ACDMPV and Hydronephrosis.** Genes (Basel). 14(3):563. doi: 10.3390/genes14030563. PMID: 36980834 (IF=4,141) (MNiSW= 100)

Paprocka J, Jezela-Stanek A, Śmigiel R, Walczak A, Mierzewska H, Kutkowska-Kaźmierczak A, Płoski R, Emich-Widera E, Steinborn B (2023) **Expanding the Knowledge of KIF1A-Dependent Disorders to a Group of Polish Patients.** Genes (Basel). 14(5):972. doi: 10.3390/genes14050972. PMID: 37239332 (IF=4,141) (MNiSW= 100)

Braun-Walicka N, Pluta A, Wolak T, Maj E, Maryniak A, Gos M, Abramowicz A, Landowska A, Obersztyn E, Bal J (2023) **Research on the Pathogenesis of Cognitive and Neurofunctional Impairments in Patients with Noonan Syndrome: The Role of Rat Sarcoma–Mitogen Activated Protein Kinase Signaling Pathway Gene Disturbances.** Genes (Basel) 14(12):2173. <https://doi.org/10.3390/genes14122173> (IF=4,141) (MNiSW= 100)