

**PUBLIKACJE:**

Oracz G, Zaród M, Ewers M, Laumen H, Gambin T, Kamiński P, Grabowska I, Drożak A, Kwiatkowski S, Wertheim-Tysarowska K, Kołodziejczyk E, Domaszewicz A, Dorożko B, Kosińska J, Głuszek S, Koziół D, Płoski R, Rosendahl J, Witt H, Drożak J, Rygiel AM (2021) **Loss of function TRPV6 variants are associated with chronic pancreatitis in nonalcoholic early-onset Polish and German patients.** *Pancreatology* 21(8): 1434-1442. (IF=3,629) (MNI<sub>SW</sub>=70)

Sobstyl M, Stapińska-Syniec A, Zaremba J, Jurek M, Kupryjaniuk A, Rylski M (2021) **Bilateral pallidal stimulation in a family with Myoclonus Dystonia Syndrome due to a mutation in sarcoglycan gene.** *Neuromodulation: Technology at the Neural Interface*. doi: 10.1111/ner.13362 (IF=4,029) (MNI<sub>SW</sub>=100)

Wernick AI, Walton RL, Soto-Beasley AI, Koga S, Heckman MG, Valentino RR, Milanowski LM, Hoffman-Zacharska D, Koziółowski D, Hassan A, Uitti RJ, Cheshire WP, Singer W, Wszolek ZK, Dickson DW, Low PA, Ross OA (2021) **Frequency of spinocerebellar ataxia mutations in patients with multiple system atrophy.** *Clin Auton Res*. 31(1):117-125 (IF= 2.968) (MNI<sub>SW</sub>=40)

Milanowski ŁM, Lindemann JA, Hoffman-Zacharska D, Soto-Beasley AI, Barcikowska M, Boczarska-Jedynak M, Deutschlander A, Kłodowska G, Dulski J, Fedoryshyn L, Friedman A, Jamrozik Z, Janik P, Karpinsky K, Koziółowski D, Krygowska-Wajs A, Jasinska-Myga B, Opala G, Potulska-Chromik A, Pulyk A, Rektorova I, Sanotsky Y, Siuda J, Stawek J, Smitowska K, Szczechowski L, Rudzinska-Bar M, Walton RL, Ross OA, Wszolek ZK (2021) **Frequency of mutations in PRKN, PINK1, and DJ1 in Patients With Early-Onset Parkinson Disease from neighboring countries in Central Europe.** *Parkinsonism and Related Disorders* 86; 48–51 (IF=3,926) (MNI<sub>SW</sub>=100)

Milanowski LM, Ross OA, Friedman A, Hoffman-Zacharska D, Gorka-Skoczylas P, Jurek M, Koziółowski D, Wszolek ZK (2021) **Genetics of Parkinson's disease in the Polish population.** *Polish Journal of Neurology and Neurosurgery* 55(3):241-252 (IF=1,7) (MNI<sub>SW</sub>=100)

Potulska-Chromik A, Jędrzejowska M, Gos M, Rosiak E, Kierdaszuk B, Maruszak A, Opuchlik A, Zekanowski C, Fichna JP (2021) **Pathogenic Mutations and Putative Phenotype-Affecting Variants in Polish Myofibrillar Myopathy Patients.** *Journal of Clinical Medicine*. 10(5):914 (IF=3.303) (MNI<sub>SW</sub>=140)

Jezela-Stanek A, Bauer A, Wertheim-Tysarowska K, Bal J, Rygiel AM, Sykut-Cegielska J (2021) **Molecular depiction of the Polish patients with classical galactosemia.** *Orphanet Journal of Rare Diseases* 16(1):239. doi: 10.1186/s13023-021-01869-3. (IF=3,52) (MNI<sub>SW</sub>=100)

Kostera-Pruszczyk A, Potulska-Chromik A, Lukawska M, Hoffman-Zacharska D Lipowska M, Olchowik B, Figlerowicz M, Figlerowicz, Kanabus K, Rosiak E (2021) **Pediatric CIDP: diagnosis and management. A single center experience.** *Front. Neurol*. Vol 12, 667378 doi: 10.3389/fneur.2021.667378 (IF=2.889) (MNI<sub>SW</sub>=100)

Klaniewska M, Jędrzejowska M, Rydzanicz M, Paprocka J, Biela M, Wolanska E, Pollak A, Debek E, Sasiadek M, Ploski R, Gos M, Smigiel R (2021) **Further delineation of neurological symptoms in young children with distal arthrogryposis, with impaired proprioception and touch caused by compound heterozygous mutation in the PIEZO2 gene.** *Front. Genet*. 12:620752 doi.org/10.3389/fgene.2021.620752 (IF=3.789) (MNI<sub>SW</sub>=100)

Jędrzejowska M, Potulska-Chromik A, Gos M, Gambin T, Dębek E, Rosiak E, Stępień A, Szymańczak R, Wojtaś W, Gielniewski B, Ciara E, Sobczyńska A, Chrzanowska K, Kostera-Pruszczyk A, Madej-Pilarczyk A (2021) **Floppy infant syndrome as a first manifestation of LMNA-related congenital muscular dystrophy.** *Eur J Paediatr Neurol* 32:115-121. (IF=3,14) (MNI<sub>SW</sub>=100)

Slezak R, Smigiel R, Obersztyn E, Pollak A, Dawidziuk M, Wiszniewski W, Bekesińska-Figatowska M, Rydzanicz M, Ploski R, Gawlinski P (2021) **Further delineation of phenotype and genotype of primary microcephaly syndrome with cortical malformations associated with compound heterozygous mutations in WDR62 gene.** *Genes (Basel)* 19;12(4):594 (IF=3,331) (MNI<sub>SW</sub>=100)

Wertheim-Tysarowska K, Oracz G, Rygiel AM (2021) **Genetic risk factors in early onset non-alcoholic pancreatitis – an update.** *Genes (Basel)* 12(5), 785 (IF=3,331) (MNI<sub>SW</sub>=100)

Bukowska-Olech E, Gawliński P, Jakubiuk-Tomaszuk A, Jędrzejowska M, Obersztyn E, Piechota M, Bielska M, Jamsheer A (2021) **The first description of monozygotic twins with craniofrontonasal syndrome discordant for the clinical phenotype but harbouring an identical nonsense variant in the EFNB1 gene.** *Orphanet Journal of Rare Diseases* 16(1):286. doi: 10.1186/s13023-021-01914-1 (IF=3,52) (MNI<sub>SW</sub>=100)

Kutkowska-Kaźmierczak AD, Boczar M, Kalka E, Castaneda J, Kłapecki J, Pietrzyk A, Barczyk A, Malinowska O, Landowska A, Gambin T, Kowalczyk K, Wisniowiecka-Kowalnik B, Smyk M, Dawidziuk M, Paczkowska M, Szyld P, Lipska-Ziętkiewicz B, Szczatuba K, Kostyk E, Runge A, Rutkowska K, Płoski R, Nowakowska B, Bal J, Obersztyn E, Gos M (2021) **Wide fontanels, delayed speech development and a hoarse voice as useful signs in the diagnosis of KBG syndrome. Clinical description of 23 cases with pathogenic variant involving the ANKRD11 gene or submicroscopic chromosomal rearrangements of 16q24.3.** *Genes (Basel)* 12(8):1257. doi.org/10.3390/genes12081257 (IF=3,7) (MNI<sub>SW</sub>=100)

Zwara A, Wertheim-Tysarowska K, Mika A (2021) **Alterations of ultra long-chain fatty acids in hereditary skin diseases – review article.** *Frontiers in Medicine* doi.org/10.3389/fmed.2021.730855 (IF=3,331) (MNI<sub>SW</sub>=70)

Johannesen KM, Liu Y, Gjerulfsen CE, Koko M, Sonnenberg L, Schubert J, Fenger ChD, Eltokhi A, Rannap M, Koch NA, Lauxmann S, Krüger J, Kegele J, Canafoglia L, Franceschetti S, Mayer T, Rebstock J, Zacher P, Ruf S, Alber M, Sterbova K, Lassuthová P, Vlckova M, Lemke JR, Krey I, Heine C, Wieczorek D, Kroell-Seger J, Lund C, Klein KM, Au PB, Rho JM, Ho AW, Masnada S, Veggiotti P, Giordano L, Accorsi P, Che H-H, Striano P, Zara F, Verhelst H, Verhoeven JS, van der Zwaag B, Harder AVE, Brilstra E, Pendziwiat M, Lebon S, Vaccarezza M, Ngoc Minh Le, Christensen J, Schmidt-Petersen MU, Grønberg S, Scherer SW, Howe J, Walid Fazeli, Howell KB, Leventer R, Stutterd C, Walsh S, Gerard M, Gerard B, Matricardi S, Bonardi CM, Sartori S, Berger A, Hoffman-Zacharska D, Mastrangelo M, Darra F, Vøllo A, Motazacker MM, Lakeman Ph, Nizon M, Betzler C, Altuzarra C, Caume R, Roubertie A, Gélisse Ph, Marini C, Guerrini R, Bilan F, Tibussek D, Koch-Hogrebe M, Perry MS, Ichikawa Sh, Dadali E, Sharkov A, Mishina I, Abramov M, Kanivets I, Korostelev S, Kutsev S, Wain KE, Eisenhauer N, Wagner M, Savatt JM, Müller-Schlüter K, Bassan H, Borovikov A, Nassogne M-C, Destrée A, Schoonjans A-S, Meuwissen M, Buzatu M, Jansen A, Scalais E, Srivastava S, Tan W-H, Olson HE, Loddenkemper T, Poduri A, Katherine L Helbig, Helbig I, Fitzgerald MP, Goldberg EM, Roser T, Borggraefe I, Brünger T, May P, Lal D, Lederer D, Rubboli G, Lesca G, Hedrich UBS, Benda J, Gardella E, Lerche H, Møller RS (2021) **Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications.** *Brain* 10.1093/brain/awab321 (IF=14,250) (MNI<sub>SW</sub>=200)

Manivannan SN, Roovers J, Smal N, Myers CT, Turkdogan D, Roelens F, Kanca O, Chung H-L, Scholz T, Hermann K, Bierhals T, Caglayan HS, Stamberger H, MAE working group of EuroEPINOMICS RES Consortium (Hoffman-Zacharska D), Mefford H, de Jonghe P, Yamamoto S, Weckhuysen S, Bellen HJ (2021) **De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies.** *Brain* doi: 10.1093/brain/awab409 (IF=14,250) (MNI<sub>SW</sub>=200)

Hernandez-Dorransoro U, Gawlinski P, Lasa-Aranzasti A, Martínez-Soroa I, Aizalde EA, Villaverde RS, Rodríguez CA, Aritzituri MS (2021) **Kosaki overgrowth syndrome due to a novel de novo PDGFRB variant** *Clinical Genetics* 10.1111/cge.14078 (IF 4,428) (MNI<sub>SW</sub>=100)

Wertheim-Tysarowska K, Szczygielski O, Seliga K, Tysarowski A, Bal J, Michalak E, Rygiel AM, Sawicka E (2021) **The retrospective molecular analysis of large or giant congenital melanocytic nevi in a cohort of Polish.** Journal of Mother and Child 25(1):19-24 (MNiSW=20)

Dawidziuk M, Kutkowska-Kaźmierczak A, Gawliński P, Wiszniewski W, Gos M, Stawiński P, Rydzanicz M, Kosińska J, Własienko P, Kordowska O, Bartnik-Głaska M, Bernaciak J, Szczatuba K, Geremek M, Dudarewicz L, Obersztyn E, Paczkowska M, Smyk M, Sobecka K, Nowakowska B (2021) **Null variants in AGRN cause lethal fetal akinesia deformation sequence.** Clin Genet. 97(4):634-638. (IF=4,438) (MNiSW=100)

Dawidziuk M, Gambin T, Bukowska-Olech E, Antczak-Marach D, Badura-Stronka M, Buda P, Budzyska E, Castaneda J, Chilarska T, Czyzyk E, Eckersdorf-Mastalerz A, Fijak-Moskal J, Gieruszczak-Bialek D, Glodek-Brzozowska E, Goszczanska-Ciuchta A, Grzeszykowska-Podymniak M, Gurda B, Jakubiuk-Tomaszuk A, Jamroz E, Janeczko M, Jedlinska-Pijanowska D, Jurek M, Karolewska D, Kazmierczak A, Kleist T, Kochanowska I, Krajewska-Walasek M, Kufel K, Kutkowska-Kaźmierczak A, Lipiec A, Maksym-Gasiorek D, Materna-Kiryłuk A, Mazurkiewicz H, Milewski M, Pavina-Guglas T, Pietrzyk A, Posmyk R, Pyrkosz A, Rudzka-Dybala M, Slezak R, Wisniewska M, Zalewska-Miszkurka Z, Szczepanik E, Obersztyn E, Bekiesinska-Figatowska M, Gawlinski P, Wiszniewski W (2021) **Exome sequencing reveals gene mutations landscape in patients with congenital microcephaly** Genes (Basel) doi.org/10.3390/genes12122014 (IF=4,096) (MNiSW=100)

Osipowicz K, Wertheim-Tysarowska K, Kwiek B, Jankowska E, Gos M, Charzewska A, Woźniak K, Kowalewski C (2021) **Bullous diseases caused by KRT1 gene mutations: from epidermolytic hyperkeratosis to a novel variant of epidermolysis bullosa simplex.** Adv Dermatol Allergol 38(6): 1032–1038 (IF=1,837) (MNiSW=70)

Jakubiak A, Szczatuba K, Badura-Stronka M, Kutkowska-Kaźmierczak A, Jakubiuk-Tomaszuk A, Chilarska T, Pilch J, Braun-Walicka N, Castaneda J, Wołyńska K, Wiśniewska M, Kugaud M, Bielecka M, Pesz K, Wierzba J, Latos-Bieleńska A, Obersztyn E, Krajewska-Walasek M, Śmigiel R (2021) **Clinical characteristics of Polish patients with molecularly confirmed Mowat-Wilson syndrome.** J. Appl Genet. 62(3):477-485. (IF=3.240) (MNiSW=140)

Bayat A, Pendziwiat M, Obersztyn E, Goldenberg P, Zacher P, Döring JH, Syrbe S, Begtrup A, Borovikov A, Sharkov A, Karasińska A, Giżewska M, Mitchell W, Morava E, Møller RS, Rubboli G (2021) **Deep-Phenotyping the Less Severe Spectrum of PIGT Deficiency and Linking the Gene to Myoclonic Atonic Seizures.** Front Genet. 11;12:663643. doi: 10.3389/fgene.2021.663643. PMID: 34046058 (IF= 3.789) (MNiSW=100)

Walczak-Sztulpa J, Wawrocka A, Stańczyk M, Pesz K, Dudarewicz L, Chrul S, Bukowska-Olech E, Wieczorek-Cichecka N, Arts HH, Oud MM, Śmigiel R, Grenda R, Obersztyn E, Chrzanowska KH, Latos-Bieleńska A (2021) **Interfamilial clinical variability in four Polish families with cranioectodermal dysplasia and identical compound heterozygous variants in WDR35** Am J Med Genet A. 185(4):1195-1203. (IF=2.802) (MNiSW=100)

Domaradzka J, Deperas M, Obersztyn E, Kucińska-Chahwan A, Brison N, Van Den Bogaert K, Roszkowski T, Kędzior M, Bartnik-Głaska M, Łuszczek A, Jakubów-Durska K, Vermeesch JR, Nowakowska BA (2021) **A placental trisomy 2 detected by NIPT evolved in a fetal small Supernumerary Marker Chromosome (sSMC)** Mol Cytogenet. 14(1):18. doi: 10.1186/s13039-021-00535-4. (IF=2,009) (MNiSW=70)

Nowakowska BA, Pankiewicz K, Nowacka U, Niemiec M, Kozłowski S, Issat T (2021) **Genetic Background of Fetal Growth Restriction.** Int J Mol Sci. 23(1):36. doi: 10.3390/ijms23010036. PMID: 35008459. (IF=5.542) (MNiSW=140)

Wielgos M, Kosinski P, Jedrzejak P, Krajewska-Walasek M, Bartnik-Głaska M, Nowakowska B, Jezela-Stanek A (2021) **How does terminal 21q22 deletion really manifest? Delineation based on prenatal diagnosis and literature review.** Taiwan J Obstet Gynecol. 60(6):1121-1125. doi: 10.1016/j.tjog.2021.09.029. PMID: 34794750. (IF=1.46) (MNiSW=40)

Kowalczyk K, Bartnik-Głaska M, Smyk M, Plaskota I, Bernaciak J, Kędzior M, Wiśniowiecka-Kowalnik B, Jakubów-Durska K, Braun-Walicka N, Barczyk A, Geremek M, Castañeda J, Kutkowska-Kaźmierczak A, Własienko P, Dębska M, Kucińska-Chahwan A, Roszkowski T, Kozłowski S, Mikulska B, Issat T, Obersztyn E, Nowakowska BA (2021) **Prenatal Diagnosis by Array Comparative Genomic Hybridization in Fetuses with Cardiac Abnormalities.** Genes (Basel). 12(12):2021. doi: 10.3390/genes12122021. PMID: 34946970; PMCID: PMC8701951. (IF=4.339) (MNI<sub>SW</sub>=100)

Kucinska-Chahwan AM, Roszkowski T, Geremek M, Paczkowska MA, Ciebiera M, Bijok J, Massalska D, Panek G, Siemion K, Nowakowska BA (2021) **Prenatal diagnosis of glutaric acidemia type 2 with the use of exome sequencing - an up-to-date review and new case report.** Ginekol Pol. 92(1):51-56. doi: 10.5603/GP.a2020.0190. Epub PMID: 33448012. (IF=0,367) (MNI<sub>SW</sub>=40)

Kucińska-Chahwan A, Roszkowski T, Nowakowska B, Geremek M, Paczkowska M, Bijok J, Massalska D (2021) **Genetic causes of the skeletal system abnormalities diagnosed by prenatal sonography with the use of exome sequencing: single institution experience.** Ultrasound Obstet Gynecol. doi: 10.1002/uog.23722. Epub ahead of print. PMID: 34198368 (IF=7.299) (MNI<sub>SW</sub>=140)

Jakubiak A, Szczatuba K, Badura-Stronka M, Kutkowska-Kaźmierczak A, Jakubiuk-Tomaszuk A, Chilarska T, Pilch J, Braun-Walicka N, Castaneda J, Wołyńska K, Wiśniewska M, Kugaudo M, Bielecka M, Pesz K, Wierzba J, Latos-Bieleńska A, Obersztyn E, Krajewska-Walasek M, Śmigiel R (2021) **Clinical characteristics of Polish patients with molecularly confirmed Mowat-Wilson syndrome.** J Appl Genet. 62(3):477-485 (IF 3.240)(MNI<sub>SW</sub>140=)

Walczak-Sztulpa J, Wawrocka A, Stańczyk M, Pesz K, Dudarewicz L, Chrul S, Bukowska-Olech E, Wieczorek-Cichecka N, Arts HH, Oud MM, Śmigiel R, Grenda R, Obersztyn E, Chrzanowska KH, Latos-Bieleńska A (2021) **Interfamilial clinical variability in four Polish families with cranioectodermal dysplasia and identical compound heterozygous variants in WDR35.** Am J Med Genet A 185(4):1195-1203 (IF 2.802)(MNI<sub>SW</sub>=100)

Hogendorf A, Zieliński M, Constantinou M, Śmigiel R, Wierzba J, Wyka K, Wędrychowicz A, Jakubiuk-Tomaszuk A, Budzyska E, Piotrowicz M, Lipska-Ziętkiewicz BS, Kaczorowska E, Cieślukowska A, Kutkowska-Kaźmierczak A, Fijak-Moskal J, Kugaudo M, Kosińska-Urbańska M, Szadkowska A, Borowiec M, Niedźwiecki M, Trzonkowski P, Młynarski W (2021) **Immune Dysregulation in Patients With Chromosome 18q Deletions-Searching for Putative Loci for Autoimmunity and Immunodeficiency.** Front Immunol. 12:742834. doi: 10.3389/fimmu.2021.742834. PMID: 34867966 (IF 7.561)(MNI<sub>SW</sub>=140)

Figura M, Geremek M, Milanowski ŁM, Meisner-Kramarz I, Duszyńska-Wąs K, Szlufik S, Różański D, Smyk Marta, Koziorowski D (2021) **Movement disorders associated with chromosomal aberrations diagnosed in adult patients.** Neurologia i Neurochirurgia Polska. 2021;55(3):300-305. doi: 10.5603/PJNNS.a2021.0038 (IF 1.621)(MNI<sub>SW</sub>=100)

Geremek M, Szklanny K **Deep Learning-Based Analysis of Face Images as a Screening Tool for Genetic Syndromes.** Sensors (Basel) 21(19):6595. doi: 10.3390/s21196595 (IF=3.576) (MNI<sub>SW</sub>=100)

Grabarczyk A, Wertheim-Tysarowska K, Bal J (2021) **Dysplazje ektodermalne – klasyfikacja i mechanizmy molekularne najczęstszych zespołów chorobowych.** Postępy Biochemii 3 (MNI<sub>SW</sub>=70)

Dawidziuk M, Kutkowska-Kaźmierczak A, Gawliński P, Wiszniewski W, Gos M, Stawiński P, Rydzanicz M, Kosińska J, Własienko P, Kordowska O, Bartnik-Głaska M, Bernaciak J, Szczatuba K, Bekiesińska-Figatowska M, Płoski R, Bal J, Rzońca-Niewczas S (2021) **The MED13L haploinsufficiency syndrome associated with de novo nonsense variant (p.Gln1981\*).** Journal of Mother and Child 30; 24(3):32-36. (MNI<sub>SW</sub>=20)

Gos M, Jędrzejowska M, Ołtarzewski M (2021) **Badania przesiewowe noworodków w kierunku rdzeniowego zaniku mięśni.** Postępy Neonatologii 3(27): 27-35 (MNI<sub>SW</sub>=20)

Rzońca-Niewczas S, Wierzbą J, Kaczorowska E, Poryszewska M, Kosińska J, Stawiński P, Płoski R, Bał J (2021) **WDR13: a novel gene implicated in non-syndromic intellectual disability**. *Genes (Basel)* 12(12):1911. doi: 10.3390/genes12121911 (IF=4,096) (MNI*SW*= 100)