

## Zoznam publikačnej činnosti

**RNDr. Ján Radvánszky, PhD.**

### **ABD Kapitoly vo vedeckých monografiách vydané v domácich vydavateľstvách**

ABD01 Kádaši, Ludevít (aut) [UKOPRBMB] (50%) - Radvánszky, Ján (aut) [UKOPRBMB] (50%): Duchenneova a Beckerova svalová dystrofia  
Lit.: 232 záz., 3 obr., 1 tab.  
In: Časté monogénne dedičné ochorenia na Slovensku. - Bratislava : VEDA, 2014. - S. 213-253 [2,99 AH]. - ISBN 978-80-224-1363-3  
Gašperíková, Daniela [rec.]  
Kovács, László [rec.]

ABD02 Radvánszky, Ján (aut) [UKOPRBMB] (100%): Myotonická dystrofia  
Lit.: 121 záz., 3 obr.  
In: Časté monogénne dedičné ochorenia na Slovensku. - Bratislava : VEDA, 2014. - S. 390-414 [1,76 AH]. - ISBN 978-80-224-1363-3  
Gašperíková, Daniela [rec.]  
Kovács, László [rec.]

ABD03 Radvánszky, Ján (aut) [UKOPRBMB] (100%): Ochorenie Charcot-Marie-Tooth typu 1A a hereditárna neuropatia s náchylnosťou na tlakovú obrnu  
Lit.: 48 záz., 3 obr.  
In: Časté monogénne dedičné ochorenia na Slovensku. - Bratislava : VEDA, 2014. - S. 415-429 [1,03 AH]. - ISBN 978-80-224-1363-3  
Gašperíková, Daniela [rec.]  
Kovács, László [rec.]

ABD04 Radvánszky, Ján (aut) [UKOPRBMB] (100%): Vybrané metódy využívané v molekulárnej diagnostike monogénne dedičných ochorení  
Lit.: 50 záz., 11 obr.  
In: Časté monogénne dedičné ochorenia na Slovensku. - Bratislava : VEDA, 2014. - S. 617-645 [1,80 AH]. - ISBN 978-80-224-1363-3  
Gašperíková, Daniela [rec.]  
Kovács, László [rec.]

### **ACB Vysokoškolské učebnice vydané v domácich vydavateľstvách**

ACB01 Radvánszky, Ján (aut) [UKOPRBMB] (50% [3,76 AH]) - Szemes, Tomáš (aut) [UKOPRBMB] (50% [3,76 AH]): Úvod do biomedicínskych aplikácií vysokoparalelného sekvenovania. - 1. vyd. - Bratislava : Centrum vedecko-technických informácií SR, 2021. - 121s. [7,52 AH] [print]  
Lit.: 8 záz.  
ISBN 978-80-89965-89-2  
učebnica pre vysoké školy  
Turňa, Ján [rec.]  
Burjanivová, Tatiana [rec.]

### **ADC Vedecké práce v zahraničných karentovaných časopisoch**

ADC01 Pálffy, Roland [UKOLF] - Gardlík, Roman [UKOLFUMB] - Hodosy, Július [UKOLF] - Behuliak, Michal [UKOLF] - Reško, P. - Radvánszky, Ján [UKOPRBMB] - Celec, Peter [UKOLFUMB]: Bacteria in gene therapy: bactofection versus alternative gene therapy  
Lit.: 40 záz.  
In: Gene Therapy. - Vol. 13, No. 2 (2006), s. 101-105. - ISSN 0969-7128

*Registrované v:*

WOS CC Web of Science Core Collection

SCOPUS SCOPUS

*Indikátor časopisu:*

IF (JCR) 2006=4,782

*Kvartil Q:*

wos-jcr -- Q1 [Biochemistry & molecular biology] -- 2006

wos-jcr -- Q1 [Biotechnology & applied microbiology] -- 2006

wos-jcr -- Q1 [Genetics & heredity] -- 2006

wos-jcr -- Q1 [Medicine, research & experimental] -- 2006

*Ohlasy (76):*

[o1] 2006 ~ Li, C.X. - Parker, A. - Menocal, E. - Xiang, S. - Borodyansky, L. - Fruehauf, J.H.: Cell. Cycle, Vol. 5, No. 18, 2006, s. 2103-2109 -- SCI ; SCOPUS

[o1] 2007 ~ Conese, M. - Boyd, A.C. - Di Gioia, S. - Auriche, C. - Ascenzioni, F.: Current Gene Therapy, Vol. 7, No. 3, 2007, s. 175-187 -- SCI ; SCOPUS

[o1] 2008 ~ Nagata, T. - Koide, Y.: Anti-infective vaccine strategies. In: Handbook of Listeria Monocytogenes. Boca Raton : CRC Press, 2008, s. 449-480 -- SCOPUS

[o1] 2008 ~ St Jean, A.T. - Zhang, M. - Forbes, N.S.: Current Opinion in Biotechnology, vol. 19, no. 5, 2008, s. 511-517 -- SCI ; SCOPUS

[o1] 2009 ~ Kubiak, K. - Nawrot, B.: Biotechnologia, no. 1, 2009, s. 132-151 -- SCOPUS

[o1] 2009 ~ Lee, C.H. - Wu, C.L. - Chen, S.H. - Shiau, A.L.: Journal of Immunotherapy, vol. 32, no. 4, 2009, s. 376-388 -- SCI ; SCOPUS

[o1] 2009 ~ Dai, M.S. - Nitcheu-Tefit, J. - Alcock, S. - Ramirez-Jimenez, F. - Chao, T.Y. - Baril, P. - Rocha, M. - Brett, S.J. - Stauss, H.J. - Vassaux, G.: Journal of Immunotherapy, vol. 32, no. 8, 2009, s. 845-855 -- SCI ; SCOPUS

[o1] 2009 ~ Bueno, S.M. - González, P.A. - Kalergis, A.M.: Current Gene Therapy, vol. 9, no. 3, 2009, s. 171-184 -- SCI ; SCOPUS

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[o1] 2010 ~ Min, J.J. - Nguyen, V.H. - Gambhir, S.S.: Nuclear Medicine and Molecular Imaging, vol. 44, no. 1, 2010, s. 15-24 -- SCOPUS

[o1] 2010 ~ Forbes, N.S.: Nature Reviews Cancer, vol. 10, no. 11, 2010, s. 784-793 -- SCI ; SCOPUS

[o1] 2010 ~ Baban, C.K. - Cronin, M. - O'Hanlon, D. - O'Sullivan, G.C. - Tangney, M.: Bioengineered Bugs, vol. 1, no. 6, 2010, s. 385-394 -- SCOPUS

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[o1] 2010 ~ Klasson, L. - Andersson, S.G.E.: BioEssays, vol. 32, no. 4, 2010, s. 288-295 -- SCI ; SCOPUS

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[o1] 2011 ~ Chang, C.H. - Cheng, W.J. - Chen, S.Y. - Kao, M.C. - Chiang, C.J. - Chao, Y.P.: Biotechnology and Bioengineering, vol. 108, no. 7, 2011, s. 1662-1672 -- SCI ; SCOPUS

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[o1] 2011 ~ Yu, H.: Applied Microbiology and Biotechnology, vol. 92, no. 6, 2011, s. 1107-1113 -- SCI ; SCOPUS

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[o1] 2011 ~ Vonka, V.: Gene Therapy: Hopes and Problems. In: Genes and Cardiovascular Function. New York : Springer, 2011, s. 7-17 -- BKCI-S

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- [o1] 2013 ~ Kwon, S.Z. - Min, J.J.: Genetically Engineered Salmonella Typhimurium for Targeted Cancer Therapy. In: Gene Therapy of Cancer: Translational Approaches from Preclinical Studies to Clinical Implementation: Third Edition. Elsevier : Amstredam, 2013, s. 443-452 -- SCOPUS
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[n1] 2022 zz ~ García-Álvarez, R. - Vallet-Regí, M.: Expert Opinion on Drug Delivery, vol. 19, no. 1, 2022, s. 103-118 -- SCI ; SCOPUS

[n1] 2022 zz ~ Jani, D. - Boyanapalli, R. - Cao, L.: Gene Therapy and Cell Therapy: Bioanalytical Challenges and Practical Solutions. In: AAPS Advances in the Pharmaceutical Sciences Series, Vol. 57. Luxemburg : Springer, 2022, S. 103-130 --SCOPUS

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[n1] 2022 zz ~ Sepich-Poore, G.D. - Guccione, C. - Laplane, L. - Pradeu, T. - Curtius, K. - Knight, R.: BioEssays, vol. 44, no. 5, 2022, art. no. 2100252 -- SCI ; SCOPUS

[n1] 2022 zz ~ Debasmitha, D. - Ghosh, S.S. - Chattopadhyay, A.: ACS Applied Bio Materials, vol. 5, no. 6, 2022, s. 2543-2548 -- SCI ; SCOPUS

[n1] 2022 zz ~ Hu, X. - Zhou, W. - Pi, R. - Zhao, X. - Wang, W.: Medicinal Research Reviews, vol. 42, no. 4, 2022, s. 1492-1517 -- SCI ; SCOPUS

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ADC02 Radvánszky, Ján (aut) [UKOPRBMB] (25%) - Resko, Peter (aut) (20%) - Surov, Milan (aut) [UKOPRBMB] (20%) - Minárik, Gabriel (aut) [UKOLF] (10%) - Ficek, Andrej (aut) [UKOPRBMB] (10%) -

Kádaši, Eudevít (aut) [UKOPRBMB] (15%): High-resolution melting analysis for genotyping of the myotonic dystrophy type 1 associated Alu insertion/deletion polymorphism

Lit. 14 zázň., 2 obr.

In: Analytical Biochemistry. - Vol. 398, No. 1 (2010), S. 126-128. - ISSN 0003-2697

*Indikátor časopisu:*

IF (JCR) 2010=3,236

*Ohlasy (12):*

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[o1] 2012 ~ Ngui, R. - Lim, Y.A.L. - Chua, K.H.: PLoS ONE, Vol. 7, No. 7, 2012, Art. No. 41996 -- SCI ; SCOPUS

[o1] 2012 ~ Er, T.-K. - Chang, J.-G.: Clinica Chimica Acta, Vol. 414, 2012, s. 197-201 -- SCI

[o1] 2014 ~ Aghaei, A.A. - Rassi, Y. - Sharifi, I. - Vatandoost, H. - Mollaie, H.R. - Oshaghi, M.A. - Abai, M.R. - Rafizadeh, S.: Asian Pacific Journal of Tropical Medicine, Vol. 7, No. 2, 2014, s. 93-96 -- SCI ; SCOPUS

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[o1] 2017 ~ Li, X.-Y. - Zhang, C. - Zhang, Q.-L. - Zhu, J.-L. - Liu, Q. - Chen, M.-W. - Yang, X.-M. - Hui, W.-L. - Cui, Y.-L.: Scientific Reports, Vol. 7, No. 1, 2017, Art. No. 8346 -- SCI ; SCOPUS

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[o1] 2019 ~ Xuhong, Y. - Sinong, Z. - Jianping, L. - Yu, C. - Juanli, Z. - Chao, Z. - Desheng, L. - Kai, H. - Yali, C. - Wenli, H.: Artificial Cells, Nanomedicine and Biotechnology, Vol. 47, No. 1, 2019, s. 636-643 -- SCI ; SCOPUS

[o1] 2019 ~ Darvishi, F.Z. - Boroumand, F. - Saadat, M.: Gene Reports, Vol. 15, Jun, 2019, Art. No. 100367 -- SCI ; SCOPUS

ADC03 Radvánszky, Ján (aut) [UKOPRBMB] (70%) - Kádaši, Eudevít (aut) [UKOPRBMB] (30%): The Expanding World of Myotonic Dystrophies: How Can They Be Detected?

Lit.: 72 zázň., 1 tab.

In: Genetic Testing and Molecular Biomarkers. - Vol. 14, No. 6 (2010), s. 733-741. - ISSN 1945-0265

*Indikátor časopisu:*

IF (JCR) 2010=0,879

*Ohlasy (4):*

[o1] 2012 ~ Van Blitterswijk, M. - DeJesus-Hernandez, M. - Rademakers, R.: Current Opinion in Neurology, Vol. 25, No. 6, 2012, s. 689-700 -- SCI ; SCOPUS

[o1] 2013 ~ Orpana, A.K. - Ho, T.H. - Alagund, K. - Ridanpää, M. - Aittomäki, K. - Stenman, J.: Journal of Molecular Diagnostics, Vol. 15, No. 1, 2013, s. 110-115 -- SCI ; SCOPUS

[o1] 2013 ~ van Blitterswijk, M. - DeJesus-Hernandez, M. - Niemantsverdriet, E. - Murray, M.E. - Heckman, M.G. - Diehl, N.N. - Brown, P.H. - Baker, M.C. - Finch, N.A. - Bauer, P.O. - Serrano, G. - Beach, T.G. - Josephs, K.A. - Knopman, D.S. - Petersen, R.C. - Boeve, B.F. - Graff-Radford, N.R. - Boylan, K.B. - Petrucelli, L. - Dickson, D.W. - Rademakers, R.: The Lancet Neurology, Vol. 12, No. 10, 2013, s. 978-988 -- SCI ; SCOPUS

[o1] 2014 ~ Cooper-Knock, J. - Shaw, P.J. - Kirby, J.: Acta Neuropathologica, Vol. 127, No. 3, 2014, s. 333-345 -- SCI ; SCOPUS

ADC04 Radvánszky, Ján (aut) [UKOPRBMB] (20%) - Bazsalovicsová, Eva (aut) [UKOPRBZO] (50%) - Králová-Hromadová, Ivica (aut) (10%) - Minárik, Gabriel (aut) [UKOLF] (10%) - Kádaši, Eudevít (aut) [UKOPRBMB] (10%): Development of high-resolution melting (HRM) analysis for population studies of Fascioloides magna (Trematoda: Fasciolidae), the giant liver fluke of ruminants

Lit.: 41 zázn., 2 obr., 3 tab.

In: Parasitology Research. - Vol. 108, No. 1 (2011), s. 201-209. - ISSN 0932-0113

*Registrované v:*

CCC Current Contents Connect

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WOS CC Web of Science Core Collection

*Indikátor časopisu:*

IF (JCR) 2011=2,149

*Kvartil Q:*

wos-jcr -- Q2 [Parasitology] -- 2011

*Ohlasy (16):*

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[o1] 2012 ~ Ngui, R. - Lim, Y.A.L. - Chua, K.H.: PLoS ONE, Vol. 7, Iss. 7, 2012, Art.No. 41996 -- SCI ; SCOPUS

[o1] 2012 ~ Kuster, C.J. - Von Elert, E.: Molecular Ecology Resources, vol. 12, no. 6, 2012, s. 1048-1057 -- SCI ; SCOPUS

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[o1] 2013 ~ Demeler, J. - Ramunke, S. - Wolken, S. - Ianiello, D. - Rinaldi, L. - Gahutu, J.B. - Cringoli, G. - van Samson-Himmelstjerna, G. - Krucken, J.: Plos One, vol. 8, no. 4, 2013, Art. No. e61285 -- SCI ; SCOPUS

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[n1] 2022 zz ~ Sindicic, M. - Bujanic, M. - Posavec, E. - Blaskovic, S. - Svetlicic, I. - Martinkovic, F. - Konjevic, D.: Comparison of mitochondrial cox1 gene in Fascioloides magna from different host species. In: Veterinarski Arhiv, Vol. 92, No. 5, 2022, s. 617-621 -- SCOPUS

ADC05 Radvánszky, Ján (aut) [UKOPRBMB] (60%) - Ficek, Andrej (aut) [UKOPRBMB] (20%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (20%): Repeat-Primed Polymerase Chain Reaction in Myotonic Dystrophy Type 2 Testing

Lit.: 13 zázn., 1 obr.

In: Genetic Testing and Molecular Biomarkers. - Vol. 15, No. 3 (2011), s. 133-136. - ISSN 1945-0265

*Indikátor časopisu:*

IF (JCR) 2011=1,110

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ADC06 Radvánszky, Ján (aut) [UKOPRBMB] (60%) - Ficek, Andrej (aut) [UKOPRBMB] (10%) - Minárik, Gabriel (aut) [UKOLF] (10%) - Pálffy, Roland (aut) (10%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (10%): Effect of Unexpected Sequence Interruptions to Conventional PCR and Repeat Primed PCR in Myotonic Dystrophy Type 1 Testing

Lit. 16 zázň., 1 obr.

In: Diagnostic Molecular Pathology. - Vol. 20, No. 1 (2011), s. 48-51. - ISSN 1052-9551

*Indikátor časopisu:*

IF (JCR) 2011=2,257

*Kvartil Q:*

wos-jcr -- Q2 [Pathology] -- 2011

*Ohlasy (27):*

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- [o1] 2012 ~ Li, Y. - Da, Y.W.: Chinese Journal of Medical Genetics, Vol. 29, Iss. 1, 2012, s. 16-18 -- SCOPUS
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- [o1] 2012 ~ Kamsteeg, E.J. - Kress, W. - Catalli, C. - Hertz, J.M. - Witsch-Baumgartner, M. - Buckley, M.F. - Van Engelen, B.G.M. - Schwartz, M. - Scheffer, H.: European Journal of Human Genetics, vol. 20, no. 12, 2012, s. 1203-1208 -- SCI ; SCOPUS
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- [o1] 2017 ~ Pešovic, J. - Peric, S. - Brkušaniin, M. - Brajuškovic, G. - Rakočević-Stojanovic, V. - Savic-Pavicevic, D.: *Neurogenetics*, Vol. 18, No. 4, 2017, s. 207-218 -- SCI ; SCOPUS
- [o1] 2018 ~ Lan, X. - Li, N. - Wan, H. - Luo, L. - Wu, Y. - Li, S. - An, Y. - Wu, B.-L.: *Journal of Genetics and Genomics*, Vol. 45, No. 10, 2018, s. 549-552 -- SCOPUS
- [o1] 2019 ~ Lian, M. - Lee, C.G. - Chong, S.S.: *Frontiers in Genetics*, Vol. 10, June, 2019, Art. No. 589 -- SCI ; SCOPUS
- [o1] 2019 ~ Leferink, M. - Wong, D.P.W. - Cai, S. - Yeo, M. - Ho, J. - Lian, M. - Kamsteeg, E.-J. - Chong, S.S. - Haer-Wigman, L. - Guan, M.: *Scientific Reports*, Vol. 9, June, 2019, Art. No. 8280 -- SCI ; SCOPUS
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- [o1] 2020 ~ Ballester-Lopez, A. - Nunez-Manchon, J. - Koehorst, E. - Linares-Pardo, I. - Almendrote, M. - Lucente, G. - Guanyabens, N. - Lopez-Osias, M. - Suarez-Mesa, A. - Hanick, S.A. - Chojnacki, J. - Lucia, A. - Pintos-Morell, G. - Coll-Canti, J. - Martinez-Pineiro, A. - Ramos-Fransi, A. - Nogales-Gadea, G.: *Three-dimensional imaging in myotonic dystrophy type 1*. In: *Neurology: Genetics*, Vol. 6, No. 4, 2020, Art. No. e484 -- SCOPUS
- [n1] 2021 zz ~ Mangin, A. - de Pontual, L. - Tsai, Y.-C. - Monteil, L. - Nizon, M. - Boisseau, P. - Mercier, S. - Ziegler, J. - Harting, J. - Heiner, C. - Gourdon, G. - Tome, S.: *Robust detection of somatic mosaicism and repeat interruptions by long-read targeted sequencing in myotonic dystrophy type 1*. In: *International Journal of Molecular Sciences*, Vol. 22, No. 5, 2021, Art. No. 2616 -- SCOPUS
- [n1] 2022 zz ~ Alfano, M. - De Antoni, L. - Centofanti, F. - Visconti, V.V. - Maestri, S. - Esposti, C.D. - Massa, R. - D'apice, M.R. - Novelli, G. - Delledonne, M. - Botta, A. - Rossato, M.: *Characterization of full-length CNBP expanded alleles in myotonic dystrophy type 2 patients by Cas9-mediated enrichment and nanopore sequencing*. In: *eLife*, Vol. 11, 2022, Art. No. e80229 -- SCOPUS
- [n1] 2022 zz ~ Peric, S. - Pesovic, J. - Savic-Pavicevic, D. - Stojanovic, V.R. - Meola, G.: *Molecular and clinical implications of variant repeats in myotonic dystrophy type 1*. In: *International Journal of Molecular Sciences*, Vol. 23, No. 1, 2022, Art. No. 354 -- SCOPUS

ADC07 Radvánszky, Ján (aut) [UKOPRBMB] (60%) - Ficek, Andrej (aut) [UKOPRBMB] (20%) - Kádaši, Ludevít [UKOPRBMB] (20%): *Upgrading molecular diagnostics of myotonic dystrophies: Multiplexing for simultaneous characterization of the DMPK and ZNF9 repeat motifs*

Lit.: 24 zázň., 2 obr.

In: *Molecular and Cellular Probes*. - Vol. 25, No. 4 (2011), s. 182-185. - ISSN 0890-8508

*Indikátor časopisu:*

IF (JCR) 2011=2,078

*Ohlasy (14):*

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[o1] 2013 ~ Bustin, S.: *International Journal of Molecular Sciences*, Vol. 14, No. 8, 2013, s. 15878-15884 -- SCI ; SCOPUS



- [o1] 2014 ~ Nakayama, T. - Nakamura, H. - Oya, Y. - Kimura, T. - Imahuku, I. - Ohno, K. - Nishino, I. - Abe, K. - Matsuura, T.: Journal of Human Genetics, Vol. 59, No. 3, 2014, s. 129-133 -- SCI ; SCOPUS
- [o1] 2014 ~ Kumar, A. - Agarwal, S. - Phadke, S.R. - Pradhan, S.: Meta Gene, Vol. 2, No. 1, 2014, s. 106-113 -- SCOPUS
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- [o1] 2015 ~ Meola, G. - Cardani, R.: Biochimica et Biophysica Acta - Molecular Basis of Disease, Vol. 1852, No. 4, 2015, s. 594-606 -- SCI ; SCOPUS
- [o1] 2017 ~ Vohánka, S.: Česká a Slovenská Neurologie a Neurochirurgie, Vol. 80, No. 3, 2017, s. 255-265 -- SCI ; SCOPUS
- [o1] 2017 ~ Yum, K. - Wang, E.T. - Kalsotra, A.: Current Opinion in Genetics and Development, Vol. 44, Jun, 2017, s. 30-37 -- SCI ; SCOPUS
- [o1] 2017 ~ Pešovic, J. - Peric, S. - Brkušaniin, M. - Brajuskovic, G. - Rakočević-Stojanovic, V. - Savic-Pavicevic, D.: Neurogenetics, Vol. 18, No. 4, 2017, s. 207-218 -- SCI ; SCOPUS
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- [o1] 2020 ~ Ballester-Lopez, A. - Linares-Pardo, I. - Koehorst, E. - Nunez-Manchon, J. - Pintos-Morell, G. - Coll-Canti, J. - Almendrote, M. - Lucente, G. - Arbex, A. - Magana, J.J. - Murillo-Melo, N.M. - Lucia, A. - Monckton, D.G. - Cumming, S.A. - Ramos-Fransi, A. - Martinez-Pineiro, A. - Nogales-Gadea, G.: The need for establishing a universal CTG sizing method in myotonic dystrophy type 1. In: Genes, Vol. 11, No. 7, 2020, Art. No. 757 -- SCOPUS
- [n1] 2022 zz ~ Soltanzadeh, P.: Myotonic Dystrophies: A Genetic Overview. In: Genes, Vol. 13, No. 2, 2022, Art. No. 367 -- SCOPUS
- [n1] 2022 zz ~ Peric, S. - Pesovic, J. - Savic-Pavicevic, D. - Stojanovic, V.R. - Meola, G.: Molecular and clinical implications of variant repeats in myotonic dystrophy type 1. In: International Journal of Molecular Sciences, Vol. 23, No. 1, 2022, Art. No. 354 -- SCOPUS

ADC08 Polák, Emil (aut) [UKOPRBMB] (25%) - Ficek, Andrej (aut) [UKOPRBMB] (15%) - Radvánszky, Ján (aut) [UKOPRBMB] (15%) - Šoltýsová, Andrea (aut) [UKOPRBMB] (15%) - Üрге, Otto (aut) (5%) - Čmelová, Eleonóra (aut) (5%) - Kantarská, Dana (aut) (5%) - Kádaši, Eudevít [UKOPRBMB] (15%): Phenylalanine hydroxylase deficiency in the Slovak population: Genotype-phenotype correlations and genotype-based predictions of BH4-responsiveness

Lit.: 40 zázň., 2 obr., 3 tab.

In: Gene. - Vol. 526, No. 2 (2013), s. 347-355. - ISSN 0378-1119

*Registrované v:*

CCC Current Contents Connect

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*Indikátor časopisu:*

IF (JCR) 2013=2,082

*Ohlasy (24):*

[o1] 2014 ~ Blau, N. - Shen, N. - Carducci, C.: Expert Review of Molecular Diagnostics, Vol. 14, No. 6, 2014, s. 655-671 -- SCI ; SCOPUS

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- [o1] 2016 ~ Blau, N.: Human Mutation, Vol. 37, No. 6, 2016, s. 508-515 -- SCI ; SCOPUS
- [o1] 2016 ~ Aldámiz-Echevarria, L. - Llarena, M. - Bueno, M.A. - Dalmau, J. - Vitoria, I. - Fernández-Marmiesse, A. - Andrade, F. - Blasco, J. - Alcalde, C. - Gil, D. - García, M.C. - González-Lamuno, D. - Ruiz, M. - Ruiz, M.A. -Pena-Quintana, L. - González, D. - Sánchez-Valverde, F. - Desviat, L.R. - Pérez, B. - Couce, M.L.: Journal of Human Genetics, Vol. 61, No. 8, 2016, s. 731-744 -- SCI ; SCOPUS
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- [o1] 2017 ~ Klaassen, K. - Stankovic, B. - Kotur, N. - Djordjevic, M. - Zukic, B. - Nikcevic, G. - Ugrin, M. - Spasovski, V. - Srzentic, S. - Pavlovic, S. - Stojiljkovic, M.: Journal of Applied Genetics, Vol. 58, No. 1, 2017, s. 79-85 -- SCI ; SCOPUS
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- [o1] 2017 ~ Pampukha, V. - Nechyporenko, M. - Livshyts, L.: Genes and Diseases, Vol. 4, No. 2, 2017, s. 108-110 -- SCI ; SCOPUS
- [o1] 2017 ~ Zhu, T. - Ye, J. - Han, L. - Qiu, W. - Zhang, H. - Liang, L. - Gu, X.: Scientific Reports, Vol. 7, No. 1, 2017, Art. No. 6762 -- SCI ; SCOPUS
- [o1] 2017 ~ Choi, R. - Lee, J. - Park, H.-D. - Park, J.E. - Kim, Y.H. - Ki, C.-S. - Lee, S.-Y. - Song, J. - Kim, J.-W. - Lee, D.H.: Journal of Pediatric Endocrinology and Metabolism, Vol. 30, No. 11, 2017, s. 1211-1218 -- SCI ; SCOPUS
- [o1] 2018 ~ Wang, Z.-W. - Jiang, S.-W. - Zhou, B.-C.: Kaohsiung Journal of Medical Sciences, Vol. 34, No. 2, 2018, s. 89-94 -- SCI ; SCOPUS
- [o1] 2018 ~ Zhang, Z. - Gao, J.-J. - Feng, Y. - Zhu, L.-L. - Yan, H. - Shi, X.-F. - Chang, A.-M. - Shi, Y. - Wang, P.: Scandinavian Journal of Clinical and Laboratory Investigation, Vol. 78, No. 3, 2018, s. 211-218 -- SCI ; SCOPUS
- [o1] 2018 ~ Vieira Neto, E. - Laranjeira, F. - Quelhas, D. - Ribeiro, I. - Seabra, A. - Mineiro, N. - M. Carvalho, L. - Lacerda, L. - G. Ribeiro, M.: Molecular Genetics and Genomic Medicine, Vol. 6, No. 4, 2018, s. 575-591 -- SCI ; SCOPUS
- [o1] 2018 ~ Zong, Y. - Liu, N. - Ma, S. - Bai, Y. - Guan, F. - Kong, X.: Gene, Vol. 668, August, 2018, s. 135-139 -- SCI ; SCOPUS
- [o1] 2019 ~ Yan, Y. - Zhang, C. - Jin, X. - Zhang, Q. - Zheng, L. - Feng, X. - Hao, S. - Gao, H. - Ma, X.: Metabolic Brain Disease, Vol. 34, No. 3, 2019, s. 733-745 -- SCOPUS
- [o1] 2019 ~ Gundorova, P. - Stepanova, A.A. - Kuznetsova, I.A. - Kutsev, S.I. - Polyakov, A.V.: PLoS ONE, Vol. 14, No. 1, 2019, Art. No. e0211048 -- SCOPUS
- [o1] 2020 ~ Yan, Y. - Jin, X. - Wang, X. - Zhang, C. - Zhang, Q. - Zheng, L. - Feng, X. - Hao, S. - Gao, H. - Ma, X.: Screening of PAH Common Mutations in Chinese Phenylketonuria Patients Using iPLEX MALDI-TOF MS. In: ACS Omega, Vol. 5, No. 4, 2020, s. 1805-1812 -- SCOPUS
- [o1] 2018 ~ Li, N. - He, C. - Li, J. - Tao, J. - Liu, Z. - Zhang, C. - Yuan, Y. - Jiang, H. - Zhu, J. - Deng, Y. - Guo, Y. - Li, Q. - Yu, P.: Scientific Reports, Vol. 8, Jul, 2018, Art. No. 11251 -- SCI ; SCOPUS
- [n1] 2021 zz ~ Oz, O. - Akbulut, E.D. - Karadag, M.E. - Gonel, A. - Koyuncu, I.: Amino acid metabolism disorders and PAH gene mutations in Southeastern Anatolia Region. In: Turkish Journal of Biochemistry, Vol. 46, No. 4, 2021, s. 387-387 --SCOPUS
- [n1] 2022 zz ~ Fishchuk, L. - Rossokha, Z. - Olkhovich, N. - Pichkur, N. - Popova, O. - Medvedieva, N. - Vershyhora, V. - Dubitska, O. - Shkurko, T. - Popovych, L. - Bondar, O. - Morozuk, I. - Onyshchenko, S. - Yevtushok, L. - Tsizh, O. -Bryl, I. - Tul, O. - Kalyinka, S. - Zinkina, I. - Matviiuk, S. - Riabova, Y. - Gorovenko, N.: The analysis of using a panel of the most common variants in the PAH gene for the newborn screening in Ukraine. In: Molecular Genetics and Metabolism Reports, Vol. 32, 2022, Art. No. 100907 -- SCOPUS

ADC09 Radvánszky, Ján (aut) [UKOPRBMB] (40%) - Surový, Milan (aut) [UKOPRBMB] (20%) - Polák, Emil (aut) [UKOPRBMB] (20%) – Kádaši, Ľudevít (aut) [UKOPRBMB] (20%): Uninterrupted CCTG tracts in the myotonic dystrophy type 2 associated locus

Lit.: 30 zázň., 2 obr., 1 tab.

In: Neuromuscular Disorders. - Vol. 23, No. 7 (2013), s. 591-598. - ISSN 0960-8966

Registrované v:

CCC Current Contents Connect

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WOS CC Web of Science Core Collection

*Indikátor časopisu:*

IF (JCR) 2013=3,134

*Ohlasy (12):*

[o1] 2013 ~ Hagerman, P.: Acta Neuropathologica, Vol. 126, No. 1, 2013, s. 1-19 -- SCI ; SCOPUS

[o1] 2015 ~ Quinn, C. - Salajegheh, M.K.: Seminars in Neurology, Vol. 35, No. 4, 2015, s. 360-368 -- SCI ; SCOPUS

[o1] 2015 ~ Guo, P. - Lam, S.L.: FEBS Letters, Vol. 589, No. 20, Part. B, 2015, s. 3058-3063 -- SCI ; SCOPUS

[o1] 2015 ~ Finsterer, J. - Rudnik-Schöneborn, S.: Fortschritte der Neurologie Psychiatrie, Vol. 83, No. 1, 2015, s. 9-17 -- SCI ; SCOPUS

[o2] 2015 ~ Mattosova, S. - Hlavata, A. - Spalek, P. - Kotysova, L. - Macekova, D. - Chandoga, J.: Bratislava Medical Journal-Bratislavské lekárske listy, Vol. 116, No. 8, 2015, s. 502-505 -- SCI ; SCOPUS

[o1] 2018 ~ Mahyera, A.S. - Schneider, T. - Halliger-Keller, B. - Schrooten, K. - Hörner, E.-M. - Rost, S. - Kress, W.: Frontiers in Neurology, Vol. 9, Jun, 2018, Art. No. 463 -- SCI ; SCOPUS

[o1] 2020 ~ Khristich, A.N. - Mirkin, S.M.: On the wrong DNA track: Molecular mechanisms of repeat-mediated genome instability. In: Journal of Biological Chemistry, Vol. 295, No. 13, 2020, s. 4134-4170 -- SCOPUS

[o1] 2020 ~ Wansink, D.G. - Gourdon, G. - van Engelen, B.G.M. - Schoser, B.: 248th ENMC International Workshop: Myotonic dystrophies: Molecular approaches for clinical purposes, framing a European molecular research network, Hoofddorp, the Netherlands, 11-13 October 2019. In: Neuromuscular Disorders, Vol. 30, No. 6, 2020, s. 521-531 -- SCOPUS

[n1] 2021 zz ~ Botta, A. - Visconti, V.V. - Fontana, L. - Bisceglia, P. - Bengala, M. - Massa, R. - Bagni, I. - Cardani, R. - Sangiuolo, F. - Meola, G. - Antonini, G. - Petrucci, A. - Pegoraro, E. - D'Apice, M.R. - Novelli, G.: A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. In: Frontiers in Genetics, Vol. 12, 2021, Art. No. 668094 -- SCOPUS

[n1] 2021 zz ~ Damen, M. - Schijvenaars, M. - Schimmel-Naber, M. - Groothuismink, J. - Coenen, M. - Tieleman, A.: Ancestral Origin of the First Indian Families with Myotonic Dystrophy Type 2. In: Journal of Neuromuscular Diseases, Vol. 8, No.4, 2021, s. 715-722 -- SCOPUS

[n1] 2022 zz ~ Belyaeva, E.O. - Lebedev, I.N.: Interloci CNV Interactions in Variability of the Phenotypes of Neurodevelopmental Disorders. In: Russian Journal of Genetics, Vol. 58, No. 10, 2022, s. 1169-1179 -- SCOPUS

[n1] 2022 zz ~ Alfano, M. - De Antoni, L. - Centofanti, F. - Visconti, V.V. - Maestri, S. - Esposti, C.D. - Massa, R. - D'apice, M.R. - Novelli, G. - Delledonne, M. - Botta, A. - Rossato, M.: Characterization of full-length CNBP expanded alleles in myotonic dystrophy type 2 patients by Cas9-mediated enrichment and nanopore sequencing. In: eLife, Vol. 11, 2022, Art. No. e80229 -- SCOPUS

ADC10 Radvánszky, Ján (aut) [KAUT] [UKOPRBMB] (20%) - Surový, Milan (aut) [UKOPRBMB] (20%) - Nagyová, Emília (aut) [UKOPRBMB] (20%) - Minárik, Gabriel (aut) [UKOLF] (20%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (20%): Comparison of different DNA binding fluorescent dyes for applications of high-resolution melting analysis

Lit.: 35 zázň., 1 obr., 2 tab.

In: Clinical Biochemistry. - Vol. 48, No. 9 (2015), s. 609-616. - ISSN 0009-9120

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2015=2,382

*Kvartil Q:*

wos-jcr -- Q2 [medical laboratory technology] -- 2015

*Ohlasy (24):*

[o1] 2016 ~ Giugliani, R. - Brusius-Facchin, A.-C. - Pasqualim, G. - Leistner-Segal, S. - Riegel, M. - Matte, U.: Expert Review of Molecular Diagnostics, Vol. 16, No. 1, 2016, s. 113-123 -- SCI ; SCOPUS

[o1] 2016 ~ Haines, A.M. - Linacre, A.: Forensic Science International, Vol. 262, May, 2016, s. 190-195 -- SCI ; SCOPUS

- [o1] 2016 ~ Luchi, N. - Capretti, P. - Pazzagli, M. - Pinzani, P.: Applied Microbiology and Biotechnology, Vol. 100, No. 12, 2016, s. 5182-5204 -- SCI ; SCOPUS
- [o1] 2016 ~ Haines, A.M. - Tobe, S.S. - Linacre, A.: BioTechniques, Vol. 61, No. 4, 2016, s. 183-189 -- SCI ; SCOPUS
- [o1] 2016 ~ Samaras, A. - Madesis, P. - Karaoglanidis, G.S.: Frontiers in Microbiology, Vol. 7, November, 2016, Art. No. 1815 -- SCI ; SCOPUS
- [o1] 2016 ~ Farrar, J.S. - Wittwer, C.T.: High-Resolution Melting Curve Analysis for Molecular Diagnostics. In: Molecular Diagnostics: Third Edition. London: Elsevier, 2016, S. 79-102 -- SCOPUS
- [o1] 2017 ~ Cheishvili, D. - Petropoulos, S. - Christiansen, S. - Szyf, M.: Targeted DNA methylation analysis methods. In: Methods in Pharmacology and Toxicology. New York: Humana Press, 2017, S. 33-50 -- SCOPUS
- [o1] 2017 ~ Wang, W. - Zijlstra, R.T. - Gänzle, M.G.: BMC Microbiology, Vol. 17, No. 1, 2017, Art. No. 114 -- SCI ; SCOPUS
- [o1] 2017 ~ Jansson, L. - Koliana, M. - Sidstedt, M. - Hedman, J.: Biotechnology Reports, Vol. 14, March, 2017, s. 34-37 -- SCOPUS
- [o1] 2017 ~ Slomka, M. - Sobalska-Kwapis, M. - Wachulec, M. - Bartosz, G. - Strapagiel, D.: International Journal of Molecular Sciences, Vol. 18, No. 11, 2017, 2316 -- SCOPUS
- [o1] 2018 ~ Pereira, L. - Gomes, S. - Barrias, S. - Fernandes, J.R. - Martins-Lopes, P.: Food Research International, Vol. 103, January 2018, s. 170-181 -- SCOPUS
- [o1] 2018 ~ Shoute, L.C.T. - Loppnow, G.R.: Physical Chemistry Chemical Physics, Vol. 20, No. 7, 2018, s. 4772-4780 -- SCI ; SCOPUS
- [o1] 2018 ~ Hou, Y.-L. - You, C.-G.: Clinical Laboratory, Vol. 64, No. 7-8, 2018, s. 1113-1119 -- SCI ; SCOPUS
- [o1] 2018 ~ Alam, M.F. - Varshney, S. - Khan, M.A. - Laskar, A.A. - Younus, H.: International Journal of Biological Macromolecules, Vol. 113, July, 2018, s. 300-308 -- SCI ; SCOPUS
- [o1] 2019 ~ Quyen, T.L. - Ngo, T.A. - Bang, D.D. - Madsen, M. - Wolff, A.: Frontiers in Microbiology, Vol. 10, 2019, Art. No. 2234 -- SCOPUS; SCI
- [o1] 2019 ~ Dong, J. - Xu, Q. - Li, C.-C. - Zhang, C.-Y.: Chemical Communications, Vol. 55, No. 17, 2019, s. 2457-2460 -- SCOPUS; SCI
- [o1] 2019 ~ Shen, C.-H.: Diagnostic molecular biology. In: Diagnostic Molecular Biology. Amsterdam : Elsevier, 2019, S. 1-451 -- SCOPUS
- [o1] 2020 ~ Bae, J.H. - Fang, J.Z. - Zhang, D.Y.: High-throughput methods for measuring DNA thermodynamics. In: Nucleic Acids Research, Vol. 48, No. 15, 2020, s. E89-E89 -- SCOPUS
- [o1] 2020 ~ Zhu, J. - Wang, J. - Cheng, K. - Chen, H. - An, R. - Zhang, Y. - Komiyama, M. - Liang, X.: Effective Characterization of DNA Ligation Kinetics by High-Resolution Melting Analysis. In: ChemBioChem, Vol. 21, No. 6, 2020, s. 785-788-- SCOPUS
- [o1] 2020 ~ Gan, C. - Huang, X. - Zhan, J. - Liu, X. - Huang, Y. - Cui, J.: Study on the interactions between B-norcholesteryl benzimidazole compounds with ct-DNA. In: Spectrochimica Acta - Part A: Molecular and Biomolecular Spectroscopy, Vol.227, 2020, Art. No. 117525 -- SCOPUS
- [n1] 2021 zz ~ Gazali, F.M. - Nuhamunada, M. - Nabilla, R. - Supriyati, E. - Hakim, M.S. - Arguni, E. - Daniwijaya, E.W. - Nuryastuti, T. - Haryana, S.M. - Wibawa, T. - Wijayanti, N.: Detection of SARS-CoV-2 spike protein D614G mutation by PCR-HRM analysis. In: Heliyon, Vol. 7, No. 9, 2021, Art. No. e07936 -- SCOPUS
- [n1] 2021 zz ~ Wan, J. - Zheng, L. - Kong, L. - Lu, Z. - Tao, Y. - Feng, Z. - Lv, F. - Meng, F. - Bie, X.: Development of a rapid detection method for real-time fluorescent quantitative PCR of Salmonella spp. and Salmonella Enteritidis in ready-to-eat fruits and vegetables. In: LWT, Vol. 149, 2021, Art. No. 111837 -- SCOPUS
- [n1] 2022 zz ~ Li, J. - Xiao, B. - Xuan, J. - Sun, Z. - Li, L.: Clinical application of multiplex nucleic acid detection technologies: advancements and future. In: Chinese Journal of Laboratory Medicine, Vol. 45, No. 4, 2022, s. 428-432 --SCOPUS
- [n1] 2022 zz ~ Dehbashi, S. - Tahmasebi, H. - Alikhani, M.Y. - Keramat, F. - Arabestani, M.R.: Optimization and development of high-resolution melting curve analysis (HRMA) assay for detection of New Delhi metallo-beta-lactamase (NDM) producing Pseudomonas aeruginosa. In: AIMS Microbiology, Vol. 8, No. 2, 2022, s. 178-192 -- SCOPUS

Mannoni, Alessandro (aut) - Santucci, Annalisa (aut) - Milucci, Lia (aut) - Sestini, Silvia (aut) - Biolcati, Gianfranco (aut) - Sorge, Fiammetta (aut) - Aurizi, Caterina (aut) - Aquaron, Robert (aut) - Alsbou, Mohammed (aut) - Marques Lourenco, Charles (aut) - Ramadevi, Kanakasabapathi (aut) - Ranganath, Lakshminarayan (aut) - Gallagher, Janes, A. (aut) - Van Kan, Christa (aut) - Hall, Anthony K. (aut) - Olsson, Birgitta (aut) - Sireau, Nicolas (aut) - Ayoob, Hana (aut) - Timmis, Oliver G. (aut) - Sang, Kim-Hanh Le Quan (aut) - Genovese, Federica (aut) - Imrich, Richard (aut) [UKOEXFL] - Rovenský, Jozef (aut) - Srinivasaraghavan, Rangan (aut) - Bharadwaj, Shruthi K. (aut) - Spiegel, Ronen (aut) - Zařková, Andrea (aut) [KAUT] : Twelve novel HGD gene variants identified in 99 alkaptonuria patients: focus on 'black bone disease' in Italy

Lit.: 38 zázň., 1 obr., 2 tab.

In: European Journal of Human Genetics. - Vol. 24, No. 1 (2016), s. 66-72. - ISSN (print) 1018-4813

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2016=4,287

*Kvartil Q:*

wos-jcr -- Q1 [biochemistry and molecular biology]; Q1 [genetics and heredity] -- 2016

*Ohlasy (6):*

[n1] 2021 zz ~ Sun, T. - Chen, Y. - Wen, Y. - Zhu, Z. - Li, M.: PremPLI: a machine learning model for predicting the effects of missense mutations on protein-ligand interactions. In: Communications Biology, Vol. 4, No. 1, 2021, Art. No. 1311 -- SCOPUS

[n1] 2021 zz ~ Bychkov, I. - Kamenets, E. - Kurkina, M. - Rychkov, G. - Ilyushkina, A. - Filatova, A. - Guseva, D. - Baydakova, G. - Nekrasov, A. - Cheblokov, A. - Skoblov, M. - Zakharova, E.: Alkaptonuria in Russia: mutational spectrum and novel variants. In: European Journal of Medical Genetics, Vol. 64, No. 4, 2021, Art. No. 104165 -- SCOPUS

[n1] 2021 zz ~ Khalil, R. - Ali, D. - Mwafi, N. - Alsaraireh, A. - Obeidat, L. - Albsoul, E. - Al, Sbou' I.: Variant Analysis of Alkaptonuria Families with Significant Founder Effect in Jordan. In: BioMed Research International, Vol. 2021, 2021, Art. No. 1515641 -- SCOPUS

[n1] 2022 zz ~ Lequeue, S. - Neuckermans, J. - Nulmans, I. - Schwaneberg, U. - Vanhaecke, T. - de Kock, J.: A robust bacterial high-throughput screening system to evaluate single nucleotide polymorphisms of human homogentisate 1,2-dioxygenase in the context of alkaptonuria. In: Scientific Reports, Vol. 12, No. 1, 2022, Art. No. 19452 -- SCOPUS

[n1] 2022 zz ~ Tao, L. - Deng, C. - Ma, M. - Zhang, Y. - Duan, J. - Li, Y. - Fang, L. - Zhou, Y. - He, X. - Wang, Y. - Wang, M. - Li, L.: A novel mutation in the homogentisate 1,2 dioxygenase gene identified in Chinese Hani pediatric patients with Alkaptonuria. In: Clinica Chimica Acta, Vol. 532, 2022, s. 164-171 -- SCOPUS

[n1] 2022 zz ~ Denisova Ekaterina, V. - Kuzin, A.V.: Ophthalmological Manifestations of Alkaptonuria. In: Oftalmologiya, Vol. 19, No. 1, 2022, s. 118-122 -- SCOPUS

ADC12 Radvánszky, Ján (aut) [UKOPRBMB] (45%) - Hýblová, Michaela (aut) [UKOPRBMBd] (7%) - Ďurovřiková, D. (aut) (7%) - Hikkelová, Martina (aut) (7%) - Fiedler, E. (aut) (6%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (7%) - Turňa, Ján (aut) [UKOPRBMB] (7%) - Minárik, Gabriel (aut) (7%) - Szemes, Tomáš (aut) [UKOPRBMB] (7%): Complex phenotypes blur conventional borders between Say-Barber-Biesecker-Young-Simpson syndrome and genitopatellar syndrome

Lit.: 15 zázň., 2 obr.

In: Clinical Genetics. - Vol. 91, No. 2, Sp. Iss. (2017), s. 339-343. - ISSN 0009-9163

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2017=3,512

*Kvartil Q:*

wos-jcr -- Q2 [genetics & heredity] -- 2017

*Ohlasy (13):*

- [o1] 2017 ~ Bashir, R.A. - Dixit, A. - Goedhart, C. - Parboosingh, J.S. - Innes, A.M. - Ferreira, P. - Hasan, S.U. - Au, P.-Y.B.: American Journal of Medical Genetics, Part A, Vol. 173, No. 10, 2017, s. 2596-2604 -- SCI ; SCOPUS
- [o1] 2017 ~ Niida, Y. - Mitani, Y. - Kuroda, M. - Yokoi, A. - Nakagawa, H. - Kato, A.: Congenital Anomalies, Vol. 57, No. 3, 2017, s. 86-88 -- SCI
- [o1] 2018 ~ Marangi, G. - Di Giacomo, M.C. - Lattante, S. - Orteschi, D. - Patrizi, S. - Doronzio, P.N. - Riviello, F.N. - Vaisfeld, A. - Frangella, S. - Zollino, M.: American Journal of Medical Genetics, Part A, Vol. 176, No. 2, 2018, s.455-459 -- SCI ; SCOPUS
- [o1] 2018 ~ Vanlerberghe, C. - Boutry, N. - Petit, F.: Clinical Genetics, Vol. 94, No. 1, 2018, s. 43-53 -- SCI ; SCOPUS
- [o1] 2020 ~ Wiesel-Motiuk, N. - Assaraf, Y.G.: The key roles of the lysine acetyltransferases KAT6A and KAT6B in physiology and pathology. In: Drug Resistance Updates, Vol. 53, 2020, Art. No. 100729 -- SCOPUS
- [o1] 2020 ~ Wu, Y. - Zhang, H. - Tang, M. - Guo, C. - Deng, A. - Li, J. - Wang, Y. - Xiao, L. - Yang, G.: High methylation of lysine acetyltransferase 6B is associated with the Cobb angle in patients with congenital scoliosis. In: Journal of Translational Medicine, Vol. 18, No. 1, 2020, Art. No. 210 -- SCOPUS
- [o1] 2020 ~ Zhu, L. - Lv, L. - Wu, D. - Shao, J.: KAT6B Genetic Variant Identified in a Short Stature Chinese Infant: A Report of Physical Growth in Clinical Spectrum of KAT6B-Related Disorders. In: Frontiers in Pediatrics, Vol. 8, 2020, Art.No. 124 -- SCOPUS
- [o1] 2020 ~ Zhang, L.X. - Lemire, G. - Gonzaga-Jauregui, C. - Molidperee, S. - Galaz-Montoya, C. - Liu, D.S. - Verloes, A. - Shillington, A.G. - Izumi, K. - Ritter, A.L. - Keena, B. - Zackai, E. - Li, D. - Bhoj, E. - Tarpinian, J.M. - Bedoukian, E. - Kukulich, M.K. - Innes, A.M. - Ediae, G.U. - Sawyer, S.L. - Nair, K.M. - Soumya, P.C. - Subbaraman, K.R. - Probst, F.J. - Bassetti, J.A. - Sutton, R.V. [et al.]: Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. In: Genetics in Medicine, Vol. 22, No. 8, 2020, s. 1338-1347 -- SCOPUS
- [o1] 2020 ~ Ferrando Meseguer, E. - Cuesta, A. - Pino, L. - Crehuá-Gaudiza, E. - Miragall, L. - Pedrosa, L. - Verdecia, A. - Mínguez, M.F.: A further patient with genitopatellar syndrome requiring multidisciplinary management. In: Clinical Dysmorphology, Vol. 29, No. 4, 2020, s. 193-196 -- SCI
- [n1] 2021 zz ~ Fallah, M.S. - Szarics, D. - Robson, C.M. - Eubanks, J.H.: Impaired Regulation of Histone Methylation and Acetylation Underlies Specific Neurodevelopmental Disorders. In: Frontiers in Genetics, Vol. 11, 2021, Art. No. 613098 --SCOPUS
- [n1] 2022 zz ~ Chen, J. - Tong, G. - Wang, Y. - Ye, F. - Shi, L. - Li, H.: Diagnosis of a child with Say-Barber-Biesecker-Young-Simpson syndrome due to variant of KAT6B gene. In: Chinese Journal of Medical Genetics, Vol. 39, No. 12, 2022, s.1370-1374 -- SCOPUS
- [n1] 2022 zz ~ Yang, Y. - Zhang, H.-M. - Huang, H.: Clinical features and the genetic analysis of KAT6B-related diseases caused by a de novo mutation of the KAT6B gene c.621+1G>A. In: Asian Journal of Surgery, Vol. 45, No. 2, 2022, s.792-794 -- SCOPUS
- [n1] 2022 zz ~ Wilson, K.D. - Porter, E.G. - Garcia, B.A.: Reprogramming of the epigenome in neurodevelopmental disorders. In: Critical Reviews in Biochemistry and Molecular Biology, Vol. 57, No. 1, 2022, s. 73-112 -- SCOPUS

ADC13 Budiš, Jaroslav (aut) [UKOVP] (20%) - Kucharík, Marcel (aut) (20%) - Ďuriš, František (aut) (5%) - Gazdarica, Juraj (aut) [UKOPRBMB] (5%) - Zrubcová, Michaela (aut) (5%) - Ficek, Andrej (aut) [UKOPRBMB] (5%) - Szemes, Tomáš (aut) [UKOPRBMB](10%) - Brejová, Bronislava (aut) [UKOMFKI] (10%) - Radvánszky, Ján (aut) [UKOPRBMB] (20%): Dante: genotyping of known complex and expanded short tandem repeats

Lit.: 35 zázn.

In: Bioinformatics. - Roč. 35, č. 8 (2019), s. 1310-1317. - ISSN (print) 1367-4803

*Registrované v:*

SCO SCOPUS

SCIE Science Citation Index Expanded

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*Indikátor časopisu:*

IF (JCR) 2019=5,61

*Kvartil Q:*

wos-jcr -- Q1 [Biotechnology & applied microbiology] -- 2019

wos-jcr -- Q1 [Biochemical research methods] -- 2019

wos-jcr -- Q1 [Mathematical & computational biology] -- 2019

*Ohlasy (6):*

[o1] 2019 ~ Kinney, N. - Kang, L. - Eckstrand, L. - Pulenthiran, A. - Samuel, P. - Anandakrishnan, R. - Varghese, R.T. - Michalak, P. - Garner, H.R.: PLoS ONE, Vol. 14, No. 12, 2019, Art. No. e0225216 -- SCI ; SCOPUS

[o1] 2020 ~ Halman, A. - Oshlack, A.: Accuracy of short tandem repeats genotyping tools in whole exome sequencing data. In: F1000Research, Vol. 9, 2020, Art. No. 200 -- SCOPUS

[o1] 2021 ~ Chen, H. - Lu, Y. - Lu, D. - Xu, S.: Y-Lineage Tracker: a high-throughput analysis framework for Y-chromosomal next-generation sequencing data. In: BMC Bioinformatics, Vol. 22, No. 1, 2021, art. no. 114 -- SCI ; SCOPUS

[o1] 2021 ~ Kinney, N. - Kang, L. - Bains, H. - Lawson, E. - Husain, M. - Husain, K. - Sandhu, I. - Shin, Y. - Carter, J.K. - Anandakrishnan, R. - Michalak, P. - Garner, H.: Ethnically biased microsatellites contribute to differential geneexpression and glutathione metabolism in Africans and Europeans. In: PLoS ONE, Vol. 16, No. 3, 2021, art. no. e0249148 -- SCI ; SCOPUS

[n1] 2021 zz ~ Huszar, T.I. - Gettings, K.B. - Vallone, P.M.: An introductory overview of open-source and commercial software options for the analysis of forensic sequencing data. In: Genes, Vol. 12, No. 11, 2021, Art. No. 1739 -- SCOPUS

[n1] 2022 zz ~ Chen, J. - Li, F. - Wang, M. - Li, J. - Marquez-Lago, T.T. - Leier, A. - Revote, J. - Li, S. - Liu, Q. - Song, J.: BigFiRSt: A Software Program Using Big Data Technique for Mining Simple Sequence Repeats From Large-Scale Sequencing Data. In: Frontiers in Big Data, Vol. 4, 2022, Art. No. 727216 -- SCOPUS

ADC14 Budiš, Jaroslav (aut) [UKOVP] (10%) - Gazdarica, Juraj (aut) [UKOPRBMB] (10%) - Radvánszky, Ján (aut) [UKOPRBMB] (10%) - Szűcs, Gábor (aut) [UKOMFKAMS] (10%) - Kucharík, Marcel (aut) (10%) - Striešková, Lucia (aut) [UKOVP] (4%) - Gazdaricová, Iveta (aut) [UKOPRBMB] (5%) - Haršanyová, Mária (aut) [UKOPRBMB] (5%) - Ďuriš, František (aut) (10%) - Minárik, Gabriel (aut) [UKOPRBMB] (4%) - Sekelská, Martina (aut) (4%) - Nagy, Balint (aut) (4%) - Turňa, Ján (aut) [UKOPRBMB] (4%) - Szemes, Tomáš (aut) [UKOPRBMB] (10%): Combining count- and length-based z-scores leads to improved predictions in non-invasive prenatal testing

Lit.: 45 zázn.

In: Bioinformatics. - Roč. 35, č. 8 (2019), s. 1284-1291. - ISSN (print) 1367-4803

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*Indikátor časopisu:*

IF (JCR) 2019=5,61

*Kvartil Q:*

wos-jcr -- Q1 [Biotechnology & applied microbiology] -- 2019

wos-jcr -- Q1 [Biochemical research methods] -- 2019

wos-jcr -- Q1 [Mathematical & computational biology] -- 2019

*Ohlasy (5):*

[o1] 2020 ~ Shi, J. - Zhang, R. - Li, J. - Zhang, R.: Size profile of cell-free DNA: A beacon guiding the practice and innovation of clinical testing. In: Theranostics, Vol. 10, No. 11, 2020, s. 4737-4748 -- SCI ; SCOPUS

[o1] 2021 ~ Zhou, L. - Zhang, B. - Liu, L. - Shi, Y. - Wang, J. - Yu, B.: The Optimal Cutoff Value of Z-scores Enhances the Judgment Accuracy of Non-invasive Prenatal Screening. In: Frontiers in Genetics, Vol. 12, 2021, Art. No. 690063 -- SCI ; SCOPUS

[n2] 2022 zz ~ Che, H. - Stanley, K. - Jatsenko, T. - Thienpont, B. - Vermeesch, J. R.: Expanded knowledge of cell-free DNA biology: potential to broaden the clinical utility. In: Extracellular Vesicles and Circulating Nucleic Acids, Vol.2022, No. 3, 2022, s. 199-217

[n1] 2022 zz ~ Wen, L. - Gao, J. Y. - Huang, L. L. - Li, D. M. - Zhong, G. S.: Non-invasive prenatal screening in southeast China: clinical application and accuracy evaluation. In: Expert Review of Molecular Diagnostics, Vol. 22, No. 8, 2022, s. 841-848 -- SCI ; SCOPUS

[n2] 2023 zz ~ Qian, Y. - Liu, Y. - Yan, K. - Xu, Y. - Sun, Y. - Gao, X. - Chen, N. - Jin, P. - Zeng, L. - Sun, L. - Yan, Q. - Dong, M.: Non-invasive Prenatal Screening for Common Fetal Aneuploidies Using Single-Molecule Sequencing. In: Laboratory Investigation, Vol. 103, No. 4, 2023, art. no. 100043

ADC15 Budiš, Jaroslav (aut) [UKOVP] (9%) - Gazdarica, Juraj (aut) [UKOPRBMB] (9%) - Radvánszky, Ján (aut) [UKOPRBMB] (9%) - Haršanyová, Mária (aut) [UKOPRBMB] (9%) - Gazdaricová, Iveta (aut) [UKOPRBMB] (8%) - Striešková, Lucia (aut) [UKOVP] (8%) - Frno, Richard (aut) (8%) - Ďuriš, František (aut) (8%) - Minárik, Gabriel (aut) (8%) - Sekelská, Martina (aut) (8%) - Nagy, Balint (aut) (8%) - Szemes, Tomáš (aut) [UKOPRBMB] (8%): Non-invasive prenatal testing as a valuable source of population specific allelic frequencies

Lit.: 28 zázn.

In: Journal of Biotechnology. - č. 299 (2019), s. 72-78. - ISSN (print) 0168-1656

*Registrované v:*

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SCIE Science Citation Index Expanded

*Indikátor časopisu:*

IF (JCR) 2019=3,503

*Kvartil Q:*

wos-jcr -- Q2 [Biotechnology & applied microbiology] -- 2019

*Ohlasy (3):*

[o1] 2020 ~ Tran, N.H. - Vo, T.B. - Nguyen, V.T. - Tran, N.-T. - Trinh, T.-H.N. - Pham, H.-A.T. - Dao, T.H.T. - Nguyen, N.M. - Van, Y.-L.T. - Tran, V.U. - Vu, H.G. - Bui, Q.-T.N. - Vo, P.-A.N. - Nguyen, H.N. - Nguyen, Q.-T.T. - Do, T.-T.T. - Lam, N.V. - Ngoc, P.C.T. - Truong, D.K. - Nguyen, H.-N. - Giang, H. - Phan, M.-D.: Genetic profiling of Vietnamese population from large-scale genomic analysis of non-invasive prenatal testing data. In: Scientific Reports, Vol. 10, No. 1, 2020, Art. No.19142 -- SCOPUS

[n1] 2021 zz ~ Morshneva, A. - Kozyulina, P. - Vashukova, E. - Tarasenko, O. - Dvoynova, N. - Chentsova, A. - Talantova, O. - Koroteev, A. - Ivanov, D. - Serebryakova, E. - Ivashchenko, T. - Sukhomyasova, A. - Maksimova, N. - Bepalova, O. -Kogan, I. - Baranov, V. - Glotov, A.: Pilot screening of cell-free mtdna in nipt: Quality control, variant calling, and haplogroup determination. In: Genes, Vol. 12, No. 5, 2021, Art. No. 743 -- SCOPUS

[n1] 2022 sk ~ Eva, R. - Zuzana, P. - Andrea, Z. - Michaela, H. - Frantisek, B. - Tomas, S. - Ludevit, K. - Jan, R.: Molecularly confirmed pontocerebellar hypoplasia in a large family from Slovakia with four severely affected children. In: Bratislava Medical Journal, Vol. 123, No. 8, 2022, s. 568-572 -- SCOPUS

ADC16 Gazdarica, Juraj (aut) [UKOPRBMB] (16%) - Hekel, Rastislav (aut) [UKOPRBMB] (12%) - Budiš, Jaroslav (aut) [UKOVP] (12%) - Kucharík, Marcel (aut) (12%) - Ďuriš, František (aut) (12%) - Radvánszky, Ján (aut) [UKOPRBMB] (12%) - Turňa, Ján (aut) [UKOPRBMB] (12%) - Szemes, Tomáš (aut) [UKOPRBMB] (12%): Combination of Fetal Fraction Estimators Based on Fragment Lengths and Fragment Counts in Non-Invasive Prenatal Testing

Lit.: 29 zázn.

In: International journal of molecular sciences. - Roč. 20, č. 16 (2019), s. [1-13], Art. No. 3959. - ISSN (online) 1422-0067

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2019=4,556

*Kvartil Q:*

wos-jcr -- Q1 [Biochemistry & molecular biology] -- 2019

wos-jcr -- Q2 [Chemistry, multidisciplinary] -- 2019

*Ohlasy (5):*

[o1] 2019 ~ Nagy, B.: International Journal of Molecular Sciences, Vol. 20, No. 22, 2019, Art. No. 5645 -- SCOPUS



[n1] 2021 zz ~ Cserhati, M.: Calculation of fetal fraction for non-invasive prenatal testing. In: BioTech, Vol. 10, No. 3, 2021, Art. No. 17 -- SCOPUS  
[n1] 2022 zz ~ Deng, C. - Liu, S.: Factors Affecting the Fetal Fraction in Non-invasive Prenatal Screening: A Review. In: Frontiers in Pediatrics, Vol. 10, 2022, Art. No. 812781 -- SCOPUS  
[n1] 2022 zz ~ Pratella, D. - Duboc, V. - Milanese, M. - Boudjarane, J. - Descombes, S. - Paquis-Flucklinger, V. - Bottini, S.: Genome Mixer and TRUST: Novel bioinformatics tools to improve reliability of Non-Invasive Prenatal Testing (NIPT) for fetal aneuploidies. In: Computational and Structural Biotechnology Journal, Vol. 20, 2022, s. 1028-1035 -- SCOPUS  
[n1] 2022 zz ~ Lu, J. - Sun, X. - Ma, X.: Two approaches for calculating female fetal DNA fraction in non-invasive prenatal testing based on size analysis of maternal DNA fragments. In: Biocell, Vol. 46, No. 1, 2022, s. 185-193 -- SCOPUS

ADC17 Pös, Ondrej (aut) [UKOPRBMB] (30%) - Budiš, Jaroslav (aut) [UKOVP] (30%) - Pös, Zuzana (aut) [UKOPRBMB] (20%) - Kucharík, Marcel (aut) (5%) - Ďuriš, František (aut) (5%) - Radvánszky, Ján (aut) [UKOPRBMB] (5%) - Szemes, Tomáš (aut) [UKOPRBMB] (5%): Identification of Structural Variation from NGS-Based Non-Invasive Prenatal Testing [elektronický dokument]

Lit.: 40 záz.

In: International journal of molecular sciences [elektronický dokument]. - Roč. 20, č. 18 (2019), s. [1-9], Art. No. 4403 [online]. - ISSN (online) 1422-0067

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*Indikátor časopisu:*

IF (JCR) 2019=4,556

*Kvartil Q:*

wos-jcr -- Q1 [Biochemistry & molecular biology] -- 2019

wos-jcr -- Q2 [Chemistry, multidisciplinary] -- 2019

*Ohlasy (4):*

[o1] 2019 ~ Nagy, B.: International Journal of Molecular Sciences, Vol. 20, No. 22, 2019, Art. No. 5645 -- SCOPUS ; SCI

[o1] 2020 ~ Pei, Y. - Hu, L. - Liu, J. - Wen, L. - Luo, X. - Lu, J. - Wei, F.: Efficiency of non-invasive prenatal testing for the detection of fetal microdeletions and microduplications in autosomal chromosomes. In: Molecular Genetics and Genomic Medicine, Vol. 8, No. 8, 2020, Art. No. e1339 -- SCOPUS

[n1] 2021 zz ~ Ali, T.M. - Mateu-Brull, E. - Balaguer, N. - Dantas, C. - Borges, H.R. - de Oliveira, M.Q.G. - Rodrigo, L. - Campos-Galindo, I. - Navarro, R. - Milan, M.: Inherited unbalanced reciprocal translocation with 3q duplication and 5p deletion in a foetus revealed by cell-free foetal DNA (cffDNA) testing: a case report. In: European Journal of Medical Research, Vol. 26, No. 1, 2021, Art. No. 64 -- SCOPUS

[n1] 2021 zz ~ Morshneva, A. - Kozyulina, P. - Vashukova, E. - Tarasenko, O. - Dvoynova, N. - Chentsova, A. - Talantova, O. - Koroteev, A. - Ivanov, D. - Serebryakova, E. - Ivashchenko, T. - Sukhomyasova, A. - Maksimova, N. - Beshpalova, O. - Kogan, I. - Baranov, V. - Glotov, A.: Pilot screening of cell-free mtDNA in NIPT: Quality control, variant calling, and haplogroup determination. In: Genes, Vol. 12, No. 5, 2021, Art. No. 743 -- SCOPUS

ADC18 Pös, Zuzana (aut) [UKOPRBMB] (30%) - Gyurászová, Marianna (aut) (5%) - Nagyová, Emília (aut) [UKOPRBMB] (5%) - Hýblová, Michaela (aut) (5%) - Haršanyová, Mária (aut) [UKOPRBMB] (5%) - Budiš, Jaroslav (aut) [UKOVP] (5%) - Hekel, Rastislav (aut) [UKOPRBMB] (5%) - Gazdarica, Juraj (aut) [UKOPRBMB] (5%) - Ďuriš, František (aut) (5%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (5%) - Szemes, Tomáš (aut) [UKOPRBMB] (5%) - Radvánszky, Ján (aut) [UKOPRBMB] (20%): On the critical evaluation and confirmation of germline sequence variants identified using massively parallel sequencing  
Lit.: 53 záz.

In: Journal of Biotechnology. - č. 298 (2019), s. 64-75. - ISSN (print) 0168-1656

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2019=3,503

*Kvartil Q:*

wos-jcr -- Q2 [Biotechnology & applied microbiology] -- 2019

ADC19 Pös, Zuzana (aut) [UKOPRBMB] (33.334%) - Radvánszky, Ján (aut) [KAUT] [UKOPRBMB] (33.333%) - Gardlík, Roman (aut) [UKOLFUMB] (33.333%): Cell-free nucleic acids and their emerging role in the pathogenesis and clinical management of inflammatory bowel disease [elektronický dokument]  
Lit.: 141 zázň.

In: International journal of molecular sciences [elektronický dokument]. - Roč. 20, č. 15 (2019), s. [1-21], Art. No. 3662 [online]. - ISSN (online) 1422-0067

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*Indikátor časopisu:*

IF (JCR) 2019=4,556

*Kvartil Q:*

wos-jcr -- Q1 [Biochemistry & molecular biology] -- 2019

wos-jcr -- Q2 [Chemistry, multidisciplinary] -- 2019

*Ohlasy (21):*

[o1] 2019 ~ Nagy, B.: International Journal of Molecular Sciences, vol. 20, no. 22, 2019, art. no. 5645 -- SCI ; SCOPUS

[o1] 2020 ~ Ocansey, D.K.W. - Zhang, L. - Wang, Y.F. - Yan, Y.M. - Qian, H. - Zhang, X. - Xu, W.R. - Mao, F.: Biological Reviews, vol. 95, no. 5, 2020, s. 1287-1307 -- SCI ; SCOPUS

[o1] 2020 ~ Liu, B.: Medical Hypotheses, vol. 142, 2020, art. no. 109812 -- SCI ; SCOPUS

[o1] 2020 ~ Wani, S. - Man, K. - Pothoulakis, C.: American Journal of Physiology-Gastrointestinal and Liver Physiology, vol. 319, no. 6, 2020, s. G646-G654 -- SCI ; SCOPUS

[o1] 2021 ~ del Fresno, C. - Sancho, D.: Current Opinion in Immunology, vol. 68, 2021, s. 34-40 -- SCI ; SCOPUS

[o1] 2021 ~ Mondelo-Macia, P. - Castro-Santos, P. - Castillo-Garcia, A. - Muínelo-Romay, L. - Diaz-Pena, R.: Journal of Personalized Medicine, vol. 11, no. 2, 2021, art. no. 151 -- SCI ; SCOPUS

[o1] 2021 ~ Kocana, C.C. - Toprak, S.F. - Sözer, S.: Cancer Genetics, vol. 252-253, 2021, s. 48-63 -- SCI ; SCOPUS

[o1] 2021 ~ Dong, W. - Liu, D. - Zhang, T. - You, Q. - Huang, F. - Wu, J.: Ecotoxicology and Environmental Safety, vol. 215, 2021, art. no. 112161 -- SCI ; SCOPUS

[o1] 2021 ~ Shao, B.Z. - Yao, Y. - Li, J.P. - Chai, N.L. - Linghu, E.Q.: Frontiers in Oncology, vol. 11, 2021, art. no. 714357 -- SCI ; SCOPUS

[o1] 2021 ~ dos Santos Ramos, A. - Viana, G.C.S. - de Macedo Brigido, M. - Almeida, J.F.: Pharmacological Research, vol. 171, 2021, art. no. 105779 -- SCI ; SCOPUS

[o1] 2021 ~ Lu, F. - Hong, Y. - Liu, L. - Wei, N. - Lin, Y. - He, J. - Shao, Y.: Experimental Cell Research, vol. 406, no. 2, 2021, art. no. 112756 -- SCI ; SCOPUS

[o1] 2021 ~ Shao, J. - Jin, Y. - Shao, C. - Fan, H. - Wang, X. - Yang, G.: Cellular and Molecular Biology Letters, vol. 26, no. 1, 2021, art. no. 36 -- SCI ; SCOPUS

[n1] 2022 zz ~ Le, A.P.H. - Huong, H.T.T.: IFMBE Proceedings, vol. 85, 2022, s. 637-656 -- SCOPUS

[n1] 2022 zz ~ Le, A.P.H. - Tran, T.T. - Cao, T.H.M. - Le, T.M. - Le, P.T. - Huong, H.T.T.: IFMBE Proceedings, vol. 85, 2022, s. 591-609 -- SCOPUS

[n1] 2022 zz ~ Li, F. - Yang, S. - Zhang, L. - Qiao, L. - Wang, L. - He, S. - Li, J. - Yang, N. - Yue, B. - Zhou, C.: Ecology and Evolution, vol. 12, no. 1, 2022, art. no. e8470 -- SCI ; SCOPUS

[n1] 2022 zz ~ Xie, B. - Du, K. - Huang, F. - Lin, Z. - Wu, L.: Frontiers in Pharmacology, vol. 12, 2022, art. no. 762362 -- SCI ; SCOPUS

[n1] 2022 zz ~ Eom, J.Y. - Choi, S.H. - Kim, H.J. - Kim, D.H. - Bae, J.H. - Kwon, G.S. - Lee, D.H. - Hwang, J.H. - Kim, D.K. - Baek, M.C. - Cho, Y.E.: International Journal of Molecular Sciences, vol. 23, no. 17, 2022, art. no. 9955 -- SCI ; SCOPUS

[n1] 2022 zz ~ Barta, B.B. - Simon, Á. - Nagy, L. - Dankó, T. - Raffay, R.E. - Petóvári, G. - Zsiros, V. - Sebestyén, A. - Sipos, F. - Múzes, G.: PLoS ONE, vol. 17, 2022, art. no. e0268217 -- SCOPUS

[n1] 2022 zz ~ Neuenfeldt, F. - Schumacher, J.C. - Grieshaber-Bouyer, R. - Habicht, J. - Schröder-Braunstein, J. - Gauss, A. - Merle, U. - Niesler, B. - Heineken, N. - Dalpke, A. - Gaida, M.M. - Giese, T. - Meuer, S. - Samstag, Y. - Wabnitz, G.: Cell Reports, vol. 39, no. 3, 2022, art. no. 110710 -- SCI ; SCOPUS  
[n1] 2022 zz ~ Che, H. - Jatsenko, T. - Lannoo, L. - Stanley, K. - Dehaspe, L. - Vancoillie, L. - Brison, N. - Parijs, I. - Van Den Bogaert, K. - Devriendt, K. - Severi, S. - De Langhe, E. - Vermeire, S. - Verstockt, B. - Van Calsteren, K. - Vermeesch, J.R.: npj Genomic Medicine, vol. 7, no. 1, 2022, art. no. 55 -- SCI ; SCOPUS  
[n1] 2022 zz ~ Zhang, C.Y. - Zhou, W. - Tan, Y.Q. - Tian, D.F. - Zhong, C.L.: Heliyon, vol. 8, no. 10, 2022, art. no. e11022 -- SCI

ADC20 Pös, Zuzana (aut) [UKOPRBMB] (16%) - Pös, Ondrej (aut) [UKOPRBMB] (16%) - Styk, Jakub (aut) [UKOLFULB] (16%) - Móczová, Angelika (aut) (14%) - Striešková, Lucia (aut) (14%) - Budiš, Jaroslav (aut) (6%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (6%) - Radvánszky, Ján (aut) [UKOPRBMB] (6%) - Szemes, Tomáš (aut) [UKOPRBMB] (6%): Technical and Methodological Aspects of Cell-Free Nucleic Acids Analyzes [elektronický dokument]

Lit.: 333 záz. n.

In: International journal of molecular sciences [elektronický dokument]. - Roč. 21, č. 22 (2020), s. [1-43], art. no. 8634 [online]. - ISSN (online) 1422-0067

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WOS CC Web of Science Core Collection

SCIE Science Citation Index Expanded

Indikátor časopisu:

IF (JCR) 2020=5,924

Kvartil Q:

wos-jcr -- Q1 [Biochemistry & molecular biology] -- 2020

wos-jcr -- Q2 [Chemistry, multidisciplinary] -- 2020

Ohlasy (15):

[o1] 2021 ~ Nasrollahzadeh, D. - Roshandel, G. - Delhomme, T.M. - Avogbe, P.H. - Foll, M. - Saidi, F. - Poustchi, H. - Sotoudeh, M. - Malekzadeh, R. - Brennan, P. - McKay, J. - Hainaut, P. - Abedi-Ardekani, B.: International Journal of Molecular Sciences, vol. 22, no. 11, 2021, art. no. 5627 -- SCI ; SCOPUS

[o1] 2021 ~ Russo, A. - Incorvaia, L. - Del Re, M. - Malapelle, U. - Capoluongo, E. - Gristina, V. - Castiglia, M. - Danesi, R. - Fassan, M. - Giuffrè, G. - Gori, S. - Marchetti, A. - Normanno, N. - Pinto, C. - Rossi, G. - Santini, D. - Sartore-Bianchi, A. - Silvestris, N. - Tagliaferri, P. - Troncone, G. - Cinieri, S. - Beretta, G.D.: ESMO Open, vol. 6, no. 3, 2021, art. no. 100164 -- SCI ; SCOPUS

[o1] 2021 ~ Krasic, J. - Abramovic, I. - Vrtaric, A. - Nikolac Gabaj, N. - Kralik-Oguic, S. - Katusic Bojanac, A. - Jezek, D. - Sincic, N.: Frontiers in Cell and Developmental Biology, vol. 9, 2021, art. no. 686149 -- SCI ; SCOPUS

[o1] 2021 ~ Kerachian, M.A. - Azghandi, M. - Mozaffari-Jovin, S. - Thierry, A.R.: Clinical Epigenetics, vol. 13, no. 1, 2021, art. no. 193 -- SCI ; SCOPUS

[n1] 2022 zz ~ Whitlock, J.H. - Soelter, T.M. - Williams, A.S. - Hardigan, A.A. - Lasseigne, B.N.: Human Cell, vol. 35, no. 1, 2022, s. 15-22 -- SCOPUS

[n1] 2022 zz ~ Haselmann, V. - Hedtke, M. - Neumaier, M.: Diagnostics, vol. 12, no. 3, 2022, art. no. 748 -- SCI ; SCOPUS

[n1] 2022 zz ~ Al Sharhan, N.A. - Messaoudi, S.A. - Babu, S.R. - Chaudhary, A.B. - Alsharm, A.A. - Alrefaei, A.F. - Kadasah, S. - Abu-Elmagd, M. - Assidi, M. - Buhmeida, A. - Carracedo, Á. - Almawi, W.Y.: Genes, vol. 13, no. 4, 2022, art. no. 590 -- SCI ; SCOPUS

[n1] 2022 zz ~ Liu, S. - Zhao, K. - Huang, M. - Zeng, M. - Deng, Y. - Li, S. - Chen, H. - Li, W. - Chen, Z.: Frontiers in Bioengineering and Biotechnology, vol. 10, 2022, art. no. 958134 -- SCI ; SCOPUS

[n1] 2022 zz ~ Birknerova, N. - Mancikova, V. - Paul, E.D. - Matyasovsky, J. - Cekan, P. - Palicka, V. - Parova, H.: Cancers, vol. 14, no. 19, 2022, art. no. 4882 -- SCI ; SCOPUS

[n1] 2022 zz ~ Haselmann, V. - Hedtke, M. - Neumaier, M.: Journal of Laboratory Medicine, vol. 46, no. 4, 2022, s. 225-236 -- SCI ; SCOPUS

[n1] 2022 zz ~ Kredátusová, A. - Procházka, V. - Papajík, T.: Transfúze a Hematologie Dnes, vol. 28, no. 3, 2022, s. 135-142 -- SCOPUS

[n1] 2022 zz ~ Paulson, V. - Konnick, E.Q. - Lockwood, C.H.: Clinics in Laboratory Medicine, vol. 42, no. 3, 2022, s. 485-496 -- SCI ; SCOPUS

[n1] 2023 zz ~ Bitenc, M. - Grebstad Tune, B. - Melheim, M. - Atneosen-Asegg, M. - Lai, X. - Rajar, P. - Solberg, R. - Baumbusch, L.O.: Assessing nuclear versus mitochondrial cell-free DNA (cfDNA) by qRT-PCR and droplet digital PCR using a piglet model of perinatal asphyxia. In: Molecular Biology Reports, vol. 50, no. 2, 2023, s. 1533-1544 -- SCI ; SCOPUS

[n1] 2023 zz ~ Škara, L. - Vodopić, T. - Pezelj, I. - Abramović, I. - Vrhovec, B. - Vrtarić, A. - Sinčić, N. - Tomas, D. - Bulimbašić, S. - Kuliš, T. - Ulamec, M.: Methylation pattern of caveolin-1 in prostate cancer as potential cfDNA biomarker. In: Bosnian Journal of Basic Medical Sciences, vol. 23, no. 1, 2023, s. 176-186 -- SCI ; SCOPUS

[n1] 2023 zz ~ Marin, A.M. - Sanchuki, H.B.S. - Namur, G.N. - Uno, M. - Zanette, D.L. - Aoki, M.N.: Circulating Cell-Free Nucleic Acids as Biomarkers for Diagnosis and Prognosis of Pancreatic Cancer. In: Biomedicines, vol. 11, no. 4, 2023, art. no. 1069 -- SCOPUS

ADC21 Pečimonová, Martina (aut) [UKOPRBMB] (11.112%) - Radvánszky, Ján (aut) [UKOPRBMB] (11.111%) - Smolák, Dávid (aut) [UKOPRBMB] (11.111%) - Budiš, Jaroslav (aut) [UKOVP] (11.111%) - Lichvár, Michal (aut) (11.111%) - Kristinova, Diana (aut) (11.111%) - Rozova, Ivica (aut) (11.111%) - Turňa, Ján (aut) [UKOPRBMB] (11.111%) - Szemes, Tomáš (aut) [UKOPRBMB] (11.111%): Admixed phenotype of NEDD4L associated periventricular nodular heterotopia : a case report

Lit.: 9 záz.

In: Medicine. - Roč. 100, č. 22 (2021), s. [1-5], art. no. e26136. - ISSN (print) 0025-7974

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*Indikátor časopisu:*

IF (JCR) 2021=1.817

*Kvartil Q:*

wos-jcr -- Q3 [Medicine, general & internal] -- 2021

*Ohlasy (1):*

[n1] 2022 zz ~ Santilli, C. - Aggarwal, A. - Dailey, C. - McClelland, C.: Ophthalmic Findings Associated with NEDD4L-related Disorder. In: Journal of AAPOS, Vol. 26, No. 3, 2022, s. 164-167 -- SCOPUS

ADC22 Pös, Ondrej (aut) [UKOPRBMB] (11.112%) - Radvánszky, Ján (aut) [UKOPRBMB] (11.111%) - Styk, Jakub (aut) [UKOLFULB] (11.111%) - Pös, Zuzana (aut) [UKOPRBMB] (11.111%) - Buglyó, Gergely (aut) (11.111%) - Kajsík, Michal (aut) [UKOVP] (11.111%) - Budiš, Jaroslav (aut) [UKOVP] (11.111%) - Nagy, Balint (aut) (11.111%) - Szemes, Tomáš (aut) [UKOPRBMB] (11.111%): Copy number variation [elektronický dokument] : methods and clinical applications

Lit.: 108 záz.

In: Applied sciences [elektronický dokument]. - Roč. 11, č. 2 (2021), s. [1-16], art. no. 819 [online]. - ISSN (online) 2076-3417

*Registrované v:*

SCIE Science Citation Index Expanded

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OA Open access

*Indikátor časopisu:*

IF (JCR) 2021=2,838

*Kvartil Q:*

wos-jcr -- Q3 [Chemistry, multidisciplinary] -- 2021

wos-jcr -- Q2 [Engineering, multidisciplinary] -- 2021

wos-jcr -- Q2 [Physics, applied] -- 2021

wos-jcr -- Q3 [Materials science, multidisciplinary] -- 2021

*Ohlasy (13):*

[o1] 2021 ~ Geoffroy, V. - Guignard, T. - Kress, A. - Gaillard, J.B. - Solli-Nowlan, T. - Schalk, A. - Gatinois, V. - Dollfus, H. - Scheidecker, S. - Muller, J.: *Nucleic Acids Research*, vol. 49, no. W1, 2021, s. W21-W28 -- SCI ; SCOPUS

[o1] 2021 ~ Kothary, A.S. - Mahendra, C. - Tan, M. - Min Tan, E.J. - Hong Yi, J.P. - Gabriella, G. - Hui Jocelyn, T.X. - Haruman, J.S. - Tan, Z. - Lee, C.K. - Lezhava, A. - Yan, B. - Irwanto, A.: *Pharmacogenomics*, vol. 22, no. 16, 2021, s.1041-1056 -- SCOPUS

[n1] 2022 zz ~ Arjmand, B. - Hamidpour, S.K. - Tayanloo-Beik, A. - Goodarzi, P. - Aghayan, H.R. - Adibi, H. - Larijani, B.: *Frontiers in Genetics*, vol. 13, 2022, art. no. 824451 -- SCI ; SCOPUS

[n1] 2022 zz ~ Andonegui-Elguera, S. - Silva-Román, G. - Pena-Martínez, E. - Taniguchi-Ponciano, K. - Vela-Patino, S. - Remba-Shapiro, I. - Gómez-Apo, E. - Espinosa-De-los-monteros, A.-L. - Portocarrero-Ortiz, L.A. - Guinto, G. -Moreno-Jimenez, S. - Chavez-Macias, L. - Saucedo, R. - Basurto-Acevedo, L. - Lopez-Felix, B. - Gonzalez-Torres, C. - Gaytan-Cervantes, J. - Ayala-Sumuano, J.T. - Burak-Leipuner, A. - Marrero-Rodríguez, D. - Mercado, M.: *International Journal of Molecular Sciences*, vol. 23, no. 9, 2022, art. no. 4861 -- SCI ; SCOPUS

[n1] 2022 zz ~ Jeffreys, S.A. - Becker, T.M. - Khan, S. - Soon, P. - Neubauer, H. - de Souza, P. - Powter, B.: *Frontiers in Endocrinology*, vol. 13, 2022, art. no. 895729 -- SCOPUS

[n1] 2022 zz ~ Workalemahu, T. - Dalton, S. - Allshouse, A. - Carey, A.Z. - Page, J.M. - Blue, N.R. - Thorsten, V. - Goldenberg, R.L. - Pinar, H. - Reddy, U.M. - Silver, R.M.: *BJOG: An International Journal of Obstetrics and Gynaecology*, vol.129, no. 13, 2022, s. 2125-2131 -- SCI ; SCOPUS

[n1] 2022 zz ~ Zelenova, M.A. - Iourov, I.Y.: *Current Bioinformatics*, vol. 17, no. 10, 2022, s. 883-887 -- SCOPUS

[n1] 2022 zz ~ Jiménez-Santos, M.J. - García-Martín, S. - Fustero-Torre, C. - Di Domenico, T. - Gómez-López, G. - Al-Shahrour, F.: *Molecular Oncology*, vol. 16, no. 21, 2022, s. 3881-3908 -- SCI ; SCOPUS

[n1] 2022 zz ~ Skowronek, D. - Pilz, R.A. - Bonde, L. - Schamuhn, O.J. - Feldmann, J.L. - Hoffjan, S. - Much, C.D. - Felbor, U. - Rath, M.: *International Journal of Molecular Sciences*, vol. 23, no. 24, 2022, art. no. 15639 -- SCI ; SCOPUS

[n1] 2022 zz ~ Bastos, G.C. - Tolezano, G.C. - Krepischi, A.C.V.: *Genes*, vol. 13, no. 12, 2022, art. no. 2285 -- SCI ; SCOPUS

[n1] 2022 zz ~ Büki, G. - Till, Á. - Zsigmond, A. - Bene, J. - Hadzsiev, K.: *Orvosi Hetilap*, vol. 163, no. 51, 2022, s. 2041-2051 -- SCI ; SCOPUS

[n1] 2023 zz ~ Divashuk, M.G. - Nikitina, E.A. - Sokolova, V.M. - Yurkina, A.I. - Kocheshkova, A.A. - Razumova, O.V. - Karlov, G.I. - Kroupin, P.Y.: qPCR as a Selective Tool for Cytogenetics. In: *Plants*, vol. 12, no. 1, 2023, art. no. 80 --SCI ; SCOPUS

[n1] 2023 zz ~ Smith, E.G. - Surm, J.M. - Macrander, J. - Simhi, A. - Amir, G. - Sachkova, M. Y. - Lewandowska, M. - Reitzel, A.M. - Moran, Y.: Micro and macroevolution of sea anemone venom phenotype. In: *Nature Communications*, vol. 14, no. 1,2023, art. no. 249 -- SCI ; SCOPUS

ADC23 Pös, Ondrej (aut) [UKOPRBMB] (14.29%) - Radvánszky, Ján (aut) [UKOPRBMB] (14.285%) - Buglyó, Gergely (aut) (14.285%) - Pös, Zuzana (aut) [UKOPRBMB] (14.285%) - Rusňáková, Diana (aut) [UKOPRBMB] (14.285%) - Nagy, Balint (aut) (14.285%) - Szemes, Tomáš (aut) [UKOPRBMB] (14.285%): DNA copy number variation : main characteristics, evolutionary significance, and pathological aspects  
Lit.: 124 záz.

In: *Biomedical Journal*. - Roč. 44, č. 5 (2021), s. 548-559. - ISSN (online) 2320-2890

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2021=7.892

*Kvartil Q:*

wos-jr -- Q1 [Biochemistry & molecular biology] -- 2021

wos-jr -- Q1 [Medicine, research & experimental] -- 2021

*Ohlasy (17):*

[n1] 2021 zz ~ Damian, A. - Ionescu, R.O. - Rodriguez de Alba, M. - Tamayo, A. - Trujillo-Tiebas, M.J. - Cotarelo-Perez, M.C. - Rodriguez, O.P. - Villaverde, C. - de la Fuente, L. - Romero, R. - Nunez-Moreno, G. -

Minguez, P. - Ayuso, C. - Corton, M.: Fine breakpoint mapping by genome sequencing reveals the first large x inversion disrupting the nhs gene in a patient with syndromic cataracts. In: *International Journal of Molecular Sciences*, Vol. 22, No. 23, 2021, Art. No. 12713 -- SCOPUS

[n1] 2021 zz ~ Kattner, A.A.: Greek gods and the double-edged sword of liver regeneration. In: *Biomedical Journal*, Vol. 44, No. 5, 2021, s. 515-520 -- SCOPUS

[n1] 2022 zz ~ Bastos, G.C. - Tolezano, G.C. - Krepischi, A.C.V.: Rare CNVs and Known Genes Linked to Macrocephaly: Review of Genomic Loci and Promising Candidate Genes. In: *Genes*, Vol. 13, No. 12, 2022, Art. No. 2285 -- SCOPUS

[n1] 2022 zz ~ Zhang, T. - Dong, J. - Jiang, H. - Zhao, Z. - Zhou, M. - Yuan, T.: CNV-PCC: An efficient method for detecting copy number variations from next-generation sequencing data. In: *Frontiers in Bioengineering and Biotechnology*, Vol.10, 2022, Art. No. 1000638 -- SCOPUS

[n1] 2022 zz ~ Dong, H. - Li, Z. - Bian, S. - Song, G. - Song, W. - Zhang, M. - Xie, H. - Zheng, S. - Yang, X. - Li, T. - Song, P.: Culture of patient-derived multicellular clusters in suspended hydrogel capsules for pre-clinical personalized drug screening. In: *Bioactive Materials*, Vol. 18, 2022, s. 164-177 -- SCOPUS

[n1] 2022 zz ~ Pokrovac, I. - Pezer, Z.: Recent advances and current challenges in population genomics of structural variation in animals and plants. In: *Frontiers in Genetics*, Vol. 13, 2022, Art. No. 1060898 -- SCOPUS

[n1] 2022 zz ~ Liu, Z. - Su, R. - Ahsan, A. - Liu, C. - Liao, X. - Tian, D. - Su, M.: Esophageal Squamous Cancer from 4NQO-Induced Mice Model: CNV Alterations. In: *International Journal of Molecular Sciences*, Vol. 23, No. 22, 2022, Art. No.14304 -- SCOPUS

[n1] 2022 zz ~ Belyaeva, E.O. - Lebedev, I.N.: Interloci CNV Interactions in Variability of the Phenotypes of Neurodevelopmental Disorders. In: *Russian Journal of Genetics*, Vol. 58, No. 10, 2022, s. 1169-1179 -- SCOPUS

[n1] 2022 zz ~ Sharaf-Eldin, W. - Rafat, K. - ElBagoury, N. - Zaki, M. - Essawi, M.: Egyptian female with 8q22.2q22.3 microdeletion syndrome. In: *Human Gene*, Vol. 33, 2022, Art. No. 201028 -- SCOPUS

[n1] 2022 zz ~ Fitzgerald, T. - Birney, E.: CNest: A novel copy number association discovery method uncovers 862 new associations from 200,629 whole-exome sequence datasets in the UK Biobank. In: *Cell Genomics*, Vol. 2, No. 8, 2022, Art. No.100167 -- SCOPUS

[n1] 2022 zz ~ Ding, J. - Huang, M. - Huang, B. - Peng, X. - Wu, G. - Peng, C. - Zhang, H. - Mao, C. - Wu, X.: Identification of a dysregulated ceRNA network modulated by copy number variation-driven lncRNAs in lung squamous cell carcinoma. In: *Environmental and Molecular Mutagenesis*, Vol. 63, No. 7, 2022, s. 351-361 -- SCOPUS

[n1] 2022 zz ~ Liu, Y. - Lei, C. - Wang, R. - Yang, D. - Yang, B. - Xu, Y. - Lu, C. - Wang, L. - Ding, S. - Guo, T. - Liu, S. - Luo, H.: Case Report: Whole-Exome Sequencing-Based Copy Number Variation Analysis Identified a Novel DRC1 Homozygous Exon Deletion in a Patient With Primary Ciliary Dyskinesia. In: *Frontiers in Genetics*, Vol. 13, 2022, Art. No. 940292 -- SCOPUS

[n1] 2022 zz ~ Rupp, B. - Owen, S. - Ball, H. - Smith, K.J. - Gunchick, V. - Keller, E.T. - Sahai, V. - Nagrath, S.: Integrated Workflow for the Label-Free Isolation and Genomic Analysis of Single Circulating Tumor Cells in Pancreatic Cancer. In: *International Journal of Molecular Sciences*, Vol. 23, No. 14, 2022, Art. No. 7852 -- SCOPUS

[n1] 2022 zz ~ Choudhuri, S.: Toxicological Implications of Biological Heterogeneity. In: *International Journal of Toxicology*, Vol. 41, No. 2, 2022, s. 132-142 -- SCOPUS

[n1] 2022 zz ~ Dolatabadian, A. - Fernando, W.G.D.: Genomic Variations and Mutational Events Associated with Plant-Pathogen Interactions. In: *Biology*, Vol. 11, No. 3, 2022, Art. No. 421 -- SCOPUS

[n1] 2022 zz ~ Attique, H. - Shah, S. - Jabeen, S. - Khan, F.G. - Khan, A. - Elaffendi, M.: Multiclass Cancer Prediction Based on Copy Number Variation Using Deep Learning. In: *Computational Intelligence and Neuroscience*, Vol. 2022, 2022, Art. No. 4742986 -- SCOPUS

[n1] 2022 zz ~ Raos, D. - Abramovic, I. - Tomic, M. - Vrtaric, A. - Kulis, T. - Coric, M. - Ulamec, M. - Bojanac, A.K. - Jezek, D. - Sincic, N.: CNV Hotspots in Testicular Seminoma Tissue and Seminal Plasma. In: *Cancers*, Vol. 14, No. 1, 2022, Art. No. 189 -- SCOPUS

ADC24 Radvánszky, Ján (aut) [UKOPRBMB] (10%) - Hýblová, Michaela (aut) (10%) - Radvanska, Eva (aut) (10%) - Špalek, Peter (aut) (10%) - Valachová, Alica (aut) (10%) - Magyarova, Gabriela (aut) (10%) - Bognár, Csaba (aut) (10%) - Polák, Emil (aut) (10%) - Szemes, Tomáš (aut) [UKOPRBMB] (10%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (10%): Characterisation of Non-Pathogenic Premutation-Range Myotonic Dystrophy Type 2 Alleles [elektronický dokument]

Lit.: 32 zázň.

In: Journal of clinical medicine [elektronický dokument]. - Roč. 10, č. 17 (2021), s. [1-12], art. no. 3934 [online]. - ISSN (online) 2077-0383

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2021=4.964

*Kvartil Q:*

wos-jcr -- Q2 [Medicine, general & internal] – 2021

### **ADD Vedecké práce v domácích karentovaných časopisoch**

ADD01 Resko, Peter (aut) (50%) - Radvánszky, Ján (aut) [UKOPRBMB] (10%) - Odnogová, Zuzana (aut) (5%) - Baldovič, Marián (aut) [UKOPRBMB] (5%) - Minárik, Gabriel (aut) [UKOLF] (5%) - Poláková, Helena (aut) (5%) - Pálffy, Roland (aut) (5%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (15%): Mutation analysis of PMP22 in Slovak patients with Charcot-Marie-Tooth disease and hereditary neuropathy with liability to pressure palsies

Lit.: 49 zázň., 1 obr., 3 tab.

In: General Physiology and Biophysics. - Vol. 30, No. 4 (2011), s. 379-388. - ISSN 0231-5882

*Registrované v:*

SCOPUS SCOPUS

*Indikátor časopisu:*

IF (JCR) 2011=1,192

*Kvartil Q:*

wos-jcr -- Q4 [Biochemistry & molecular biology] -- 2011

wos-jcr -- Q4 [Biophysics] -- 2011

wos-jcr -- Q4 [Physiology] -- 2011

*Ohlasy (2):*

[o1] 2015 ~ Adoukonou, T.A. - Vallat, J.-M. - Mathis, S.: African Journal of Neurological Sciences, Vol. 34, No. 1, 2015, s. 72-84 -- SCI ; SCOPUS

[o1] 2016 ~ Asadchuk, T.V. - Rumiantseva, N.V. - Naumchik, I.V. - Likhachev, S.A. - Pleshko, I.V. - Shalkevich, L.V. - Jevneronok, I.V. - Kachan, J.P.: Zhurnal Nevrologii i Psihiatrii imeni S.S. Korsakova, Vol. 2016, No. 1, 2016, s. 64-69 -- SCOPUS

ADD02 Surový, Milan (aut) [UKOPRBMB] (40%) - Šoltýsová, Andrea (aut) [UKOPRBMB] (20%) - Kolníková, Miriam (aut) [UKOLFKDN] (10%) - Sýkora, Pavol (aut) [UKOLF] (5%) - Ilenčíková, Denisa (aut) (5%) - Ficek, Andrej (aut) [UKOPRBMB] (10%) - Radvánszky, Ján (aut) [UKOPRBMB] (5%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (5%): Novel SCN1A variants in Dravet syndrome and evaluating a wide approach of patient selection

Lit.: 46 zázň., 1 obr., 2 tab.

In: General Physiology and Biophysics. - Roč. 35, č. 3 (2016), s. 333-342. - ISSN 0231-5882

*Registrované v:*

WOS CC Web of Science Core Collection

SCOPUS SCOPUS

*Indikátor časopisu:*

IF (JCR) 2016=1,170

*Kvartil Q:*

wos-jcr -- Q4 [Biochemistry & molecular biology] -- 2016

wos-jcr -- Q4 [Biophysics] -- 2016

wos-jcr -- Q4 [Physiology] -- 2016

*Ohlasy (3):*

[o1] 2017 ~ Lamar, T. - Vanoye, C.G. - Calhoun, J. - Wong, J.C. - Dutton, S.B.B. - Jorge, B.S. - Velinov, M. - Escayg, A. - Kearney, J.A.: Neurobiology of Disease, Vol. 102, June, 2017, s. 38-48 -- SCI ; SCOPUS

[o1] 2021 ~ Ademuwagun, I.A. - Rotimi, S.O. - Syrbe, S. - Ajamma, Y.U. - Adebisi, E.: *Frontiers in Neurology*, vol. 12, 2021, art. no. 600050 -- SCI ; SCOPUS

[n1] 2022 zz ~ Sullivan, J. - Deighton, A.M. - Vila, M.C. - Szabo, S.M. - Maru, B. - Gofshteyn, J.S. - James, E.S. - Rico, S. - Zuberi, S.M.: *Epilepsy and Behavior*, vol. 130, 2022, art. no. 108661 -- SCI ; SCOPUS

### **ADM Vedecké práce v zahraničných časopisoch registrovaných v databázach Web of Science alebo SCOPUS**

ADM01 Kucharík, Marcel (aut) [UKOVP] (50%) - Gnip, Andrej (aut) (5%) - Hýblová, Michaela (aut) (5%) - Budiš, Jaroslav (aut) [UKOVP] (5%) - Striešková, Lucia (aut) (5%) - Haršanyová, Mária (aut) (5%) - Pös, Ondrej (aut) [UKOPRBMB] (5%) - Pös, Zuzana (aut) [UKOPRBMB] (5%) - Radvánszky, Ján (aut) [UKOPRBMB] (5%) - Minárik, Gabriel (aut) (5%) - Szemes, Tomáš (aut) [UKOPRBMB] (5%): Non-invasive prenatal testing (NIPT) by low coverage genomic sequencing: Detection limits of screened chromosomal microdeletions [elektronický dokument]

Lit.: 30 zázn.

In: *PLoS One* [elektronický dokument]. - Roč. 15, č. 8 (2020), s. [1-15], art. no. e0238245 [online]. - ISSN (online) 1932-6203

*Registrované v:*

SCO SCOPUS

WOS CC Web of Science Core Collection

*Indikátor časopisu:*

IF (JCR) 2020=3,240

*Kvartil Q:*

wos-jcr -- Q2 [Multidisciplinary sciences] -- 2020

*Ohlasy (6):*

[n1] 2021 zz ~ Paluoja, P. - Teder, H. - Ardeshirdavani, A. - Bayindir, B. - Vermeesch, J. - Salumets, A. - Krjutskov, K. - Palta, P.: Systematic evaluation of NIPT aneuploidy detection software tools with clinically validated NIPT samples. In: *PLoS Computational Biology*, Vol. 17, No. 12, 2021, Art. No. e1009684 -- SCOPUS

[n1] 2021 zz ~ Shi, J. - Tan, P. - Li, J. - Zhang, R.: Application analysis of non invasive prenatal testing for fetal chromosome copy number variations in Chinese laboratories. In: *National Medical Journal of China*, Vol. 101, No. 15, 2021, s.1088-1092 -- SCOPUS

[n1] 2022 zz ~ Xiang, J. - Peng, Z.: Applications of Non invasive Prenatal Testing for Subchromosomal Copy Number Variations Using Cell-Free DNA. In: *Clinics in Laboratory Medicine*, Vol. 42, No. 4, 2022, s. 613-625 -- SCOPUS

[n1] 2022 zz ~ Simonini, C. - Hoopmann, M. - Kagan, K.O. - Schroder, T. - Gembruch, U. - Geipel, A.: Prenatal sonographic findings in confirmed cases of Wolf-Hirschhorn syndrome. In: *BMC Pregnancy and Childbirth*, Vol. 22, No. 1, 2022, Art. No.327 -- SCOPUS

[n1] 2022 zz ~ Zaninovic, L. - Baskovic, M. - Jezek, D. - Katusic, Bojanac A.: Validity and Utility of Non-Invasive Prenatal Testing for Copy Number Variations and Microdeletions: A Systematic Review. In: *Journal of Clinical Medicine*, Vol. 11, No. 12, 2022, Art. No. 3350 -- SCOPUS

[n1] 2022 zz ~ Pratella, D. - Duboc, V. - Milanese, M. - Boudjarane, J. - Descombes, S. - Paquis-Flucklinger, V. - Bottini, S.: Genome Mixer and TRUST: Novel bioinformatics tools to improve reliability of Non-Invasive Prenatal Testing (NIPT) for fetal aneuploidies. In: *Computational and Structural Biotechnology Journal*, Vol. 20, 2022, s. 1028-1035 -- SCOPUS

ADM02 Forgáčová, Natália (aut) [UKOPRBMB] (20%) - Gazdarica, Juraj (aut) (20%) - Budiš, Jaroslav (aut) [UKOVP] (20%) - Radvánszky, Ján (aut) [UKOPRBMB] (20%) - Szemes, Tomáš (aut) [UKOPRBMB] (20%): Repurposing non-invasive prenatal testing data: Population study of single nucleotide variants associated with Colorectal Cancer and Lynch Syndrome [elektronický dokument]

Lit.: 75 zázn.

In: *Oncology Letters* [elektronický dokument]. - Roč. 22, č. 5 (2021), s. [1-14], art. no. 779 [print]. - ISSN (print) 1792-1074

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2021=3.111

*Kvartil Q:*

wos-jcr -- Q3 [Oncology] -- 2021

ADM03 Marônek, Martin (aut) [UKOLFUMB] (7.154%) - Gromová, Barbora (aut) [UKOLFUMB] (7.142%) - Lipták, Róbert (aut) [UKOLFFYU] (7.142%) - Konečná, Barbora (aut) [UKOLFUMB] (7.142%) - Pastorek, Michal (aut) [UKOLFUMB] (7.142%) - Čechová, Barbora (aut) (7.142%) - Haršanyová, Mária (aut) [UKOPRBMB] (7.142%) - Budiš, Jaroslav (aut) [UKOVP] (7.142%) - Smolák, Dávid (aut) [UKOPRBMB] (7.142%) - Radvánszky, Ján (aut) [UKOPRBMB] (7.142%) - Szemes, Tomáš (aut) [UKOPRBMB] (7.142%) - Harsányiová, Jana (aut) [UKOLJ261] (7.142%) - Kráľová Trančíková, Alžbeta (aut) [UKOLJ110] (7.142%) - Gardlík, Roman (aut) [KAUT] [UKOLFUMB] (7.142%): Extracellular DNA correlates with intestinal inflammation in chemically induced colitis in mice [elektronický dokument]

Lit.: 48 zázn.

In: Cells [elektronický dokument]. - Roč. 10, č. 1 (2021), s. [1-19], art. no. 81 [online]. - ISSN (online) 2073-4409

*Registrované v:*

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*Indikátor časopisu:*

IF (JCR) 2021=7.666

*Kvartil Q:*

wos-jcr -- Q2 [Cell biology] -- 2021

*Ohlasy (12):*

[o1] 2021 ~ Karami Fath, M. - Aakbari Oryani, M. - Ramezani, A. - Barjoie Mojarad, F. - Khalesi, B. - Delazar, S. - Anjomrooz, M. - Taghizadeh, A. - Taghizadeh, S. - Payandeh, Z. - Pourzardosht, N.: Critical Reviews in Oncology/Hematology, vol. 166, 2021, art. no. 103477 -- SCI ; SCOPUS

[o1] 2021 ~ Khan, U. - Chowdhury, S. - Billah, M.M. - Islam, K.M.D. - Thorlaciuss, H. - Rahman, M.: International Journal of Molecular Sciences, vol. 22, no. 14, 2021, art. no. 7260 -- SCI ; SCOPUS

[o1] 2021 ~ Penate Medina, T. - Kolb, J.P. - Hüttmann, G. - Huber, R. - Penate Medina, O. - Ha, L. - Ulloa, P. - Larsen, N. - Ferrari, A. - Rafecas, M. - Ellrichmann, M. - Pravdivtseva, M.S. - Anikeeva, M. - Humbert, J. - Both, M. - Hundt, J.E. - Hövener, J.B.: Frontiers in Immunology, vol. 12, 2021, art. no. 69222 -- SCI ; SCOPUS

[o1] 2021 ~ Bruggeman, Y. - Sodr , F.M.C. - Buitinga, M. - Mathieu, C. - Overbergh, L. - Kracht, M.J.L.: Expert Opinion on Therapeutic Targets, vol. 25, no. 4, 2021, s. 269-281 -- SCI ; SCOPUS

[n1] 2022 zz ~ Ke, X.X. - Hu, T. - Jiang, M.Z.: Faseb Journal, vol. 36, no. 1, 2022, art. no. e22029 -- SCI ; SCOPUS

[n1] 2022 zz ~ Chen, F. - Liu, Y. - Shi, Y. - Zhang, J. - Liu, X. - Liu, Z. - Lv, J. - Leng, Y.: Gut Pathogens, vol. 14, no. 1, 2022, art. no. 27 -- SCI ; SCOPUS

[n1] 2022 zz ~ Dhawan, U.K. - Margraf, A. - Lech, M. - Subramanian, M.: Journal of Cellular and Molecular Medicine, vol. 26, no. 20, 2022, s. 5267-5276 -- SCI ; SCOPUS

[n1] 2022 zz ~ Muzes, G. - Bohusn  Barta, B. - Szab , O. - Horgas, V. - Sipos, F.: Biomedicines, vol. 10, no. 11, 2022, art. no. 2853 -- SCI ; SCOPUS

[n1] 2022 zz ~ Che, H. - Jatsenko, T. - Lannoo, L. - Stanley, K. - Dehaspe, L. - Vancoillie, L. - Brison, N. - Parijs, I. - Van Den Bogaert, K. - Devriendt, K. - Severi, S. - De Langhe, E.: npj Genomic Medicine, vol. 7, no. 1, 2022, art. no.55 -- SCI ; SCOPUS

[n1] 2022 zz ~ Arosa, L. - Camba-G mez, M. - Conde-Aranda, J.: Gastrointestinal Disorders, vol. 4, no. 4, 2022, s. 263-276 -- SCI ; SCOPUS

[n1] 2023 zz ~ Saez, A. - Herrero-Fernandez, B. - Gomez-Bris, R. - Sanchez-Martinez, H. - Gonzalez-Granado, J.M.: Pathophysiology of Inflammatory Bowel Disease: Innate Immune System. In: Internatioal Journal of Molecular Sciences, vol. 24, no.2, 2023, art. no. 1526 -- SCI ; SCOPUS

[n1] 2023 zz ~ Song, Y.H. - Wang, Z.J. - Kang, L. - He, Z.X. - Zhao, S.B. - Fang, X. - Li, Z.S. - Wang, S.L. - Bai, Y.: PADs and NETs in digestive system: From physiology to pathology. In: Frontiers in Immunology, vol. 14, 2023, art. no.1077041 -- SCI ; SCOPUS

ADM04 Hekel, Rastislav (aut) [UKOPRBMB] (16.67%) - Budiš, Jaroslav (aut) [UKOVP] (16.666%) - Kucharík, Marcel (aut) (16.666%) - Radvánszky, Ján (aut) [UKOPRBMB] (16.666%) - Pös, Zuzana (aut) [UKOPRBMB] (16.666%) - Szemes, Tomáš (aut) [UKOPRBMB](16.666%): Privacy preserving storage of sequenced genomic data [elektronický dokument]  
Lit.: 33 záz. n.  
In: BMC Genomics [elektronický dokument]. - Roč. 22, č. 1 (2021), s. [1-14], art. no. 712 [online]. - ISSN (online) 1471-2164  
*Registrované v:*  
SCO SCOPUS  
WOS CC Web of Science Core Collection  
*Indikátor časopisu:*  
IF (JCR) 2021=4.558  
*Kvartil Q:*  
wos-jcr -- Q2 [Biotechnology & applied microbiology] -- 2021  
wos-jcr -- Q2 [Genetics & heredity] -- 2021

#### **ADN Vedecké práce v domácich časopisoch registrovaných v databázach Web of Science alebo SCOPUS**

ADN01 Nagyová, Emília (aut) [KAUT] [UKOPRBMB] (12%) - Radvánszky, Ján (aut) [UKOPRBMB] (11%) - Hýblová, Michaela (aut) (11%) - Šimovičová, Veronika (aut) (11%) - Goncalvesová, Eva (aut) [UKOLFKK] (11%) - Asselbergs, Folkert W. (aut) (11%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (11%) - Szemes, Tomáš (aut) [UKOPRBMB] (11%) - Minárik, Gabriel (aut) [UKOPRBMB] (11%): Targeted next-generation sequencing in Slovak cardiomyopathy patients  
Lit.: 39 záz. n.  
In: Bratislava medical journal. - Roč. 120, č. 1 (2019), s. 46-51. - ISSN (print) 0006-9248  
*Registrované v:*  
SCO SCOPUS  
WOS CC Web of Science Core Collection  
*Indikátor časopisu:*  
IF (JCR) 2019=1,200  
*Kvartil Q:*  
wos-jcr -- Q3 [Medicine, general & internal] -- 2019  
*Ohlasy (3):*  
[o1] 2020 ~ Herrera-Rodríguez, D.L. - Totomoch-Serra, A. - Rosas-Madrigal, S. - Luna-Limón, C. - Marroquín-Ramírez, D. - Carnevale, A. - Rosendo-Gutiérrez, R. - Villarreal-Molina, M.T. - Márquez-Murillo, M.F.: Archivos de Cardiología de Mexico, vol. 90, no. 1, 2020, s. 58-68 -- SCOPUS  
[o1] 2020 ~ Darwish, R.K. - Haghighi, A. - Seliem, Z.S. - El-Saiedi, S.A. - Radwan, N.H. - El-Gayar, D.F. - Elfeel, N.S. - Abouelhoda, M. - Mehaney, D.A.: Cardiology in the Young, vol. 30, no. 12, 2020, s. 1910-1916 -- SCI ; SCOPUS  
[n1] 2022 zz ~ Sepp, R., Hategan, L., Csányi, B., Borbás, J., Tringer, A., Pálinkás, E.D., Nagy, V., Takács, H., Latinovics, D., Nyolczas, N., Pálinkás, A., Faludi, R., Rábai, M., Szabó, G.T., Czuriga, D., Balogh, L., Halmosi, R., Borbély, A., Habon, T., Hegedűs, Z., Nagy, I.: Diagnostics, vol. 12, no. 5, 2022, art. no. 1132 -- SCI ; SCOPUS

#### **AEC Vedecké práce v zahraničných recenzovaných vedeckých zborníkoch, monografiách**

AEC01 Zaťková, Andrea (aut) (70%) - Sedláčková, Tatiana (aut) (10%) - Radvánszky, Ján (aut) [UKOPRBMB] (5%) - Poláková, Helena (aut) (4%) - Némethová, Martina (aut) [UKOPRSV] (4%) - Aquaron, Robert (aut) (1%) - Dursun, Ismail (aut) (1%) - Usher, Jeannette L. (aut) (1%) - Kádaši, Ľudevít (aut) [UKOPRBMB] (4%): Identification of 11 Novel Homogentisate 1,2 Dioxygenase Variants in Alkaptonuria Patients and Establishment of a Novel LOVD-Based HGD Mutation Database  
Lit.: 29 záz. n., 2 obr., 2 tab.  
In: JIMD Reports - Case and Research Reports, 2012/1. Book Series: JIMD Reports, Vol. 4. - Berlin : Springer, 2013. - S. 55-65. - ISSN 2192-8304. - ISBN 978-3-642-25752-0  
*Registrované v:*  
WOS CC Web of Science Core Collection

## SCOPUS SCOPUS

*Ohlasy (19):*

- [o1] 2013 ~ Braconi, D. - Millucci, L. - Ghezzi, L. - Santucci, A.: Expert Review of Proteomics, Vol. 10, No. 6, 2013, s. 521-535 -- SCI ; SCOPUS
- [o1] 2017 ~ Albanaz, A.T.S. - Rodrigues, C.H.M. - Pires, D.E.V. - Ascher, D.B.: Expert Opinion on Drug Discovery, Vol. 12, No. 6, 2017, s. 553-563 -- SCI
- [o1] 2017 ~ Damarla, N. - Linga, P. - Goyal, M. - Tadisina, S.R. - Reddy, G.S. - Bommiseti, H.: Indian Journal of Ophthalmology, Vol. 65, No. 6, 2017, Art. No. 518 -- SCI
- [o1] 2018 ~ Kilavuz, S. - Bulut, F.D. - Kor, D. - Yilmaz, B.S. - Basaran, S. - Sarpel, T. - Mungan, N.O.: Journal of Pediatric Research, Vol. 5, No. 1, 2018, s. 7-11 -- SCI
- [o1] 2019 ~ Hughes, J.H. - Liu, K. - Plagge, A. - Wilson, P.J.M. - Sutherland, H. - Norman, B.P. - Hughes, A.T. - Keenan, C.M. - Milan, A.M. - Sakai, T. - Ranganath, L.R. - Gallagher, J.A. - Bou-Gharios, G.: Human Molecular Genetics, Vol. 28, No. 23, 2019, s. 3928-3939 -- SCOPUS ; SCI
- [o1] 2020 ~ Bernini, A. - Galderisi, S. - Spiga, O. - Amarabom, C.O. - Santucci, A.: Transient pockets as mediators of gas molecules routes inside proteins: The case study of dioxygen pathway in homogentisate 1,2-dioxygenase and its implication in Alkaptonuria development. In: Computational Biology and Chemistry, Vol. 88, 2020, Art. No. 107356 -- SCOPUS
- [o1] 2020 ~ Danda, S. - Mohan, S. - Devaraj, P. - Dutta, A.K. - Nampoothiri, S. - Yesodharan, D. - Phadke, S.R. - Jalan, A.B. - Thangaraj, K. - Verma, I.C. - Danda, D. - Jebaraj, I.: Founder effects of the homogentisate 1,2-dioxygenase (HGD) gene in a gypsy population and mutation spectrum in the gene among alkaptonuria patients from India. In: Clinical Rheumatology, Vol. 39, No. 9, 2020, s. 2743-2749 -- SCOPUS
- [o1] 2020 ~ Hughes, J.H. - Bou-Gharios, G. - Ranganath, L.R. - Gallagher, J.A.: The contribution of mouse models in the rare disease alkaptonuria. In: Drug Discovery Today: Disease Models, Vol. 31, 2020, s. 37-43 -- SCOPUS
- [o1] 2020 ~ Akbaba, A.I. - Ozgul, R.K. - Dursun, A.: Presentation of 14 alkaptonuria patients from Turkey. In: Journal of Pediatric Endocrinology and Metabolism, Vol. 33, No. 2, 2020, s. 289-294 -- SCOPUS
- [o1] 2020 ~ Dai, J. - Yu, X. - Han, Y. - Chai, L. - Liao, Y. - Zhong, P. - Xie, R. - Sun, X. - Huang, Q. - Wang, J. - Yin, Z. - Zhang, Y. - Lv, Z. - Jia, C.: TMT-labeling proteomics of papillary thyroid carcinoma reveal invasive biomarkers. In: Journal of Cancer, Vol. 11, No. 20, 2020, s. 6122-6132 -- SCOPUS
- [n1] 2021 zz ~ Bernini, A. - Petricci, E. - Atrei, A. - Baratto, M.C. - Manetti, F. - Santucci, A.: A molecular spectroscopy approach for the investigation of early phase ochronotic pigment development in Alkaptonuria. In: Scientific Reports, Vol. 11, No. 1, 2021, Art. No. 22562 -- SCOPUS
- [n1] 2021 zz ~ Lai, C.-Y. - Tsai, I.-J. - Chiu, P.-C. - Ascher, D.B. - Chien, Y.-H. - Huang, Y.-H. - Lin, Y.-L. - Hwu, W.-L. - Lee, N.-C.: A novel deep intronic variant strongly associates with Alkaptonuria. In: npj Genomic Medicine, Vol. 6, No. 1, 2021, Art. No. 89 -- SCOPUS
- [n1] 2021 zz ~ Mwafi, N. - Alasmar, A. - Al-Momani, M. - Alazaydeh, S. - Alajoulin, O. - Alsalem, M. - Kalbouneh, H.: Alkaptonuria with extensive ochronotic degeneration of the Achilles tendon and its surgical treatment: A case report and literature review. In: Asian Biomedicine, Vol. 15, No. 3, 2021, s. 129-136 -- SCOPUS
- [n1] 2021 zz ~ Kisa, P.T. - Gunduz, M. - Dorum, S. - Uzun, O.U. - Cakar, N.E. - Yildirim, G.K. - Erdol, S. - Hismi, B.O. - Tugsal, H.Y. - Ucar, U. - Gorukmez, O. - Gulden, Z.A. - Kucukongar, A. - Bulbul, S. - Sari, I. - Arslan, N.: Alkaptonuria in Turkey: Clinical and molecular characteristics of 66 patients. In: European Journal of Medical Genetics, Vol. 64, No. 5, 2021, Art. No. 104197 -- SCOPUS
- [n1] 2021 zz ~ Khalil, R. - Ali, D. - Mwafi, N. - Alsaraireh, A. - Obeidat, L. - Albsoul, E. - Al, Sbou' I.: Variant Analysis of Alkaptonuria Families with Significant Founder Effect in Jordan. In: BioMed Research International, Vol. 2021, 2021, Art. No. 1515641 -- SCOPUS
- [n1] 2022 zz ~ Grasso, D. - Geminiani, M. - Galderisi, S. - Iacomelli, G. - Peruzzi, L. - Marzocchi, B. - Santucci, A. - Bernini, A.: Untargeted NMR Metabolomics Reveals Alternative Biomarkers and Pathways in Alkaptonuria. In: International Journal of Molecular Sciences, Vol. 23, No. 24, 2022, Art. No. 15805 -- SCOPUS
- [n1] 2022 zz ~ Tao, L. - Deng, C. - Ma, M. - Zhang, Y. - Duan, J. - Li, Y. - Fang, L. - Zhou, Y. - He, X. - Wang, Y. - Wang, M. - Li, L.: A novel mutation in the homogentisate 1,2 dioxygenase gene identified in Chinese Hani pediatric patients with Alkaptonuria. In: Clinica Chimica Acta, Vol. 532, 2022, s. 164-171 -- SCOPUS
- [n1] 2022 zz ~ Karmakar, M. - Cicaloni, V. - Rodrigues, C.H.M. - Spiga, O. - Santucci, A. - Ascher, D.B.: HGDDiscovery: An online tool providing functional and phenotypic information on novel variants of

homogentisate 1,2- dioxigenase. In: Current Research in Structural Biology, Vol. 4, 2022, s. 271-277 -- SCOPUS

[n1] 2022 zz ~ Nagalakshmi, V. - Lavanya, J. - Bhavya, B. - Riya, V. - Venugopal, B. - Sai, Ramesh A.: In-silico Profiling of Deleterious Non Synonymous SNPs of Homogentisate 1, 2 Dioxygenase (HGD) Gene for Early Diagnosis of Alkaptonuria .In: Research Journal of Pharmacy and Technology, Vol. 15, No. 9, 2022, s. 3898-3904 -- SCOPUS

### V3 Vedecký výstup publikačnej činnosti z časopisu

V301 Gažiová, Michaela (aut) (8.337%) - Sládeček, Tomáš (aut) (8.333%) - Pös, Ondrej (aut) [UKOPRBMB] (8.333%) - Števkó, M. (aut) (8.333%) - Krampf, Werner (aut) [UKOPRBMB] (8.333%) - Pös, Zuzana (aut) [UKOPRBMB] (8.333%) - Hekel, Rastislav (aut) [UKOPRBMB] (8.333%) - Hlavačka, Mário (aut) (8.333%) - Kucharík, M. (aut) (8.333%) - Radvánszky, Ján (aut) [UKOPRBMB] (8.333%) - Budiš, Jaroslav (aut) [UKOVP] (8.333%) - Szemes, Tomáš (aut) [UKOPRBMB] (8.333%): Automated prediction of the clinical impact of structural copy number variations [elektronický dokument]

Lit.: 44 záz.

In: Scientific reports [elektronický dokument]. - Roč. 12, č. 1 (2022), s. [1-15], art. no. 555 [online]. - ISSN (online) 2045-2322

článok

*Registrované v:*

SCO SCOPUS

CCC Current Content Connect

WOS CC Web of Science Core Collection

*Indikátor časopisu:*

IF (JCR) 2021=4.997

Nordic List Level (Norwegian Register for Scientific Journals, Series and Publishers) 2022=1

*Kvartil Q:*

wos-jcr -- Q2 [Multidisciplinary sciences] -- 2021

V302 Radvánska, Eva (aut) (12.5%) - Pös, Zuzana (aut) (12.5%) - Zatlková, Andrea (aut) (12.5%) - Hýblová, Michaela (aut) (12.5%) - Bauer, František (aut) (12.5%) - Szemes, Tomáš (aut) [UKOPRBMB] (12.5%) - Kádaši, Eudevit (aut) [UKOPRBMB] (12.5%) - Radvánszky, Ján (aut) [UKOPRBMB] (12.5%): Molecularly confirmed pontocerebellar hypoplasia in a large family from Slovakia with four severely affected children  
Lit.: 22 záz.

In: Bratislava medical journal. - Roč. 123, č. 8 (2022), s. 568-572. - ISSN (print) 0006-9248

článok

*Registrované v:*

WOS CC Web of Science Core Collection

SCO SCOPUS

SCIE Science Citation Index Expanded

*Indikátor časopisu:*

IF (JCR) 2021=1.564

*Kvartil Q:*

wos-jcr -- Q4 [Medicine, general & internal] -- 2021

V303 Styk, Jakub (aut) [KAUT] [UKOLFULB] (30%) - Hrnčiar, Matej (aut) (10%) - Pös, Ondrej (aut) [UKOVP] (10%) - Bokorová, Silvia (aut) [UKOVP] (10%) - Klimová, Daniela (aut) [UKOLFULB] (10%) - Repiská, Vanda (aut) [UKOLFULB] (10%) - Radvánszky, Ján (aut) [UKOPRBMB] (10%) - Szemes, Tomáš (aut) [UKOPRBMB] (10%): Instability krátkých tandemových repetícií v onkológii: = Instability of short tandem repeats in oncology: the potential of liquid biopsy : potenciál tekutej biopsie  
Lit.: 31 záz.

In: NewsLab. - Roč. 13, č. 2 (2022), s. 83-86. - ISSN (print) 1338-9661

článok

- V304 Forgáčová, Natália (aut) [KAUT] [UKOPRBMB] (7.143%) - Holešová, Zuzana (aut) (7.143%) - Hekel, Rastislav (aut) (7.143%) - Sedláčková, Tatiana (aut) [UKOVP] (7.143%) - Pös, Zuzana (aut) [UKOVP] (7.143%) - Krivošíková, Lucia (aut) [UKOLFUPA] (7.143%) - Janega, Pavol (aut) [UKOLFUPA] (7.143%) - Mikuš Kuracinová, Kristína (aut) [UKOLFUPA] (7.143%) - Babál, Pavel (aut) [UKOLFUPA] (7.143%) - Radvak, Peter (aut) (7.143%) - Radvánszky, Ján (aut) [UKOPRBMB] (7.143%) - Gazdarica, Juraj (aut) (7.142%) - Budiš, Jaroslav (aut) [UKOVP] (7.143%) - Szemes, Tomáš (aut) [UKOPRBMB] (7.142%): Evaluation and limitations of different approaches among COVID-19 fatal cases using whole-exome sequencing data [elektronický dokument]  
Lit.: 65 záz. n.  
In: BMC Genomics [elektronický dokument]. - Roč. 24, č. 1 (2023), s. [1-11], art. no. 12 [online]. - ISSN (online) 1471-2164  
článok  
*Registrované v:*  
SCO SCOPUS  
OA Open access  
WOS CC Web of Science Core Collection  
*Indikátor časopisu:*  
IF (JCR) 2021=4.558  
*Kvartil Q:*  
wos-jcr -- Q2 [Biotechnology & applied microbiology] -- 2021  
wos-jcr -- Q2 [Genetics & heredity] -- 2021
- V305 Pös, Zuzana (aut) [UKOVP] (12.5%) - Khedr, Milad (aut) (12.5%) - Radvánszky, Ján (aut) [UKOPRBMB] (12.5%) - Penesová, Adela (aut) (12.5%) - Hekel, Rastislav (aut) (12.5%) - Szemes, Tomáš (aut) [UKOPRBMB] (12.5%) - Ranganath, Lakshminarayan R.(aut) (12.5%) - Zaťková, Andrea (aut) (12.5%): APOC3 and ABCA1 variants in unusual combined hypolipidaemia showing premature peripheral vascular disease  
Lit.: 38 záz. n.  
In: Bratislava medical journal. - Roč. 124, č. 5 (2023), s. 351-355. - ISSN (print) 0006-9248  
článok  
*Registrované v:*  
SCO SCOPUS  
*Indikátor časopisu:*  
IF (JCR) 2021=1.564  
Nordic List Level (Norwegian Register for Scientific Journals, Series and Publishers) 2023=1  
*Kvartil Q:*  
wos-jcr -- Q4 [Medicine, general & internal] -- 2021
- V306 Styk, Jakub (aut) [KAUT] [UKOLFULB] (30%) - Pös, Zuzana (aut) [UKOVP] (7%) - Pös, Ondrej (aut) [UKOVP] (7%) - Radvánszky, Ján (aut) [UKOPRBMB] (7%) - Hrčková Turňová, Evelína (aut) [UKOVP] (7%) - Buglyó, Gergely (aut) (7%) - Klimová, Daniela (aut) [UKOLFULB] (7%) - Budiš, Jaroslav (aut) [UKOVP] (7%) - Repiská, Vanda (aut) [UKOLFULB] (7%) - Nagy, Balint (aut) (7%) - Szemes, Tomáš (aut) [UKOPRBMB] (7%): Microsatellite instability assessment is instrumental for predictive, preventive and personalised medicine: status quo and outlook  
Lit.: 192 záz. n.  
In: EPMA Journal. - Roč. 14, č. 1 (2023), s. 143-165. - ISSN (print) 1878-5077  
článok  
*Registrované v:*  
SCO SCOPUS  
WOS CC Web of Science Core Collection  
CCC Current Content Connect  
OA Open access  
SCIE Science Citation Index Expanded  
*Indikátor časopisu:*  
IF (JCR) 2021=8.836  
AIS (JCR) 2021=0.978

*Kvartil Q:*

wos-jcr -- Q1 [Medicine, research & experimental] -- 2021

**Štatistika kategórií (Záznamov spolu: 131):**

ABD Kapitoly vo vedeckých monografiách vydané v domácich vydavateľstvách (4)  
ACB Vysokoškolské učebnice vydané v domácich vydavateľstvách (1)  
ADC Vedecké práce v zahraničných karentovaných časopisoch (24)  
ADD Vedecké práce v domácich karentovaných časopisoch (2)  
ADM Vedecké práce v zahraničných časopisoch registrovaných v databázach Web of Science alebo SCOPUS (4)  
ADN Vedecké práce v domácich časopisoch registrovaných v databázach Web of Science alebo SCOPUS (1)  
AEC Vedecké práce v zahraničných recenzovaných vedeckých zborníkoch, monografiách (1)  
AFD Publikované príspevky na domácich vedeckých konferenciách (10)  
AFG Abstrakty príspevkov zo zahraničných vedeckých konferencií (28)  
AFH Abstrakty príspevkov z domácich vedeckých konferencií (13)  
AFK Postery zo zahraničných konferencií (7)  
BDF Odborné práce v ostatných domácich časopisoch (1)  
BFA Abstrakty odborných prác zo zahraničných podujatí (konferencie, ...) (3) 58  
BFB Abstrakty odborných prác z domácich podujatí (konferencie, ...) (9)  
EDJ Prehľadové práce, odborné práce, preklady noriem; odborné preklady v časopisoch, zborníkoch (1)  
GHG Práce zverejnené spôsobom umožňujúcim hromadný prístup (1)  
V2 Vedecký výstup publikačnej činnosti ako časť editovanej knihy alebo zborníka (3)  
V3 Vedecký výstup publikačnej činnosti z časopisu (9)  
O2 Odborný výstup publikačnej činnosti ako časť knižnej publikácie alebo zborníka (9)

**Štatistika ohlasov (370):**

[o1] Citácie v zahraničných publikáciách registrované v citačných indexoch (244)  
[o2] Citácie v domácich publikáciách registrované v citačných indexoch (3)  
[n1] Citácia v publikácii registrovaná v citačných indexoch (121)  
[n2] Citácia v publikácii vrátane citácie v publikácii registrovanej v iných databázach okrem citačných indexov (2)

Dňa 8. 8. 2023

Vypracovala: Butková