

Vrijeme izvoza: 04.04.2025. 18:51:09

Repositorij: repositorij.kbsplit.hr

Ukupan broj zapisa na URL-u: 53

Broj izvezenih zapisa: 53

Naslov	URL	Autori	Naslov izvornika
The Applied Genomics Development Strategy by the Croatian Academy of Sciences and Arts paves the way for the future development of applied genomics in Croatia		Sedlic, Filip; Sertić, Jadranka; Markotić, Alemka; Primorac, Dragan; Slavica, Anita; Zibar, Lada; Vlahoviček, Kristian; Kušec, Vesna; Barić, Ivo; Paar, Vladimir; Borovečki, Fran; Žmak, Ljiljana; Kurolt, Ivan-Christian; Canki-Klain, Nina; Roksandić, Sunčana; Rinčić, Iva; Jurić, Hrvoje; Škaro, Vedrana; Marjanović, Damir; Projić, Petar; Primorac, Damir; Starčević, Antonio; Vujaklija, Dušica; Šikić, Mile; Križanović, Krešimir; Gamulin, Stjepan	
Deoxyguanosine kinase deficiency: natural history and liver transplant outcome		Manzoni, Eleonora; Carli, Sara; Gaignard, Pauline; Schlieben, Lea Dewi; Hirano, Michio; Ronchi, Dario; Gonzales, Emmanuel; Shimura, Masaru; Murayama, Kei; Okazaki, Yasushi; Barić, Ivo; Petković Ramadža, Danijela; Karall, Daniela; Mayr, Johannes; Martinelli, Diego; La Morgia, Chiara; Primiano, Guido; Santer, René; Servidei, Serenella; Bris, Céline; Cano, Aline; Furlan, Francesca; Gasperini, Serena; Laborde, Nolwenn; Lamperti, Costanza; Lenz, Dominic; Mancuso, Michelangelo; Montano, Vincenzo; Menni, Francesca; Musumeci, Olimpia; Nesbitt, Victoria; Procopio, Elena; Rouzier, Cécile; Staufner, Christian; Taanman, Jan-Willem; Tal, Galit; Ticci, Chiara; Cordelli, Duccio Maria; Carelli, Valerio; Procaccio, Vincent; Prokisch, Holger; Garone, Caterina	
Impact of genetic and non-genetic factors on phenotypic diversity in NBAS-associated disease		Hammann, Nicole; Lenz, Dominic; Barić, Ivo; Crushell, Ellen; Vici, Carlo Dionisi; Distelmaier, Felix; Feillet, Francois; Freisinger, Peter; Hempel, Maja; Khoreva, Anna L.; Laass, Martin W.; Lacassie, Yves; Lainka, Elke; Larson-Nath, Catherine; Li, Zhongdie; Lipiński, Patryk; Lurz, Eberhard; Mégarbané, André; Nobre, Susana; Olivieri, Giorgia; Peters, Bianca; Prontera, Paolo; Schlieben, Lea D.; Seroogy, Christine M.; Sobacchi, Cristina; Suzuki, Shigeru; Tran, Christel; Vockley, Jerry; Wang, Jian-She; Wagner, Matias; Prokisch, Holger; Garbade, Sven F.; Kölker, Stefan; Hoffmann, Georg F.; Staufner, Christian	

Brain function in classic galactosemia, a galactosemia network (GalNet) members review	Panis, Bianca; Vos, E. Naomi; Barić, Ivo; Bosch, Annet M.; Brouwers, Martijn C. G. J.; Burlina, Alberto; Cassiman, David; Coman, David J.; Couce, María L.; Das, Anibh M.; Demirbas, Didem; Empain, Aurélie; Gautschi, Matthias; Grafakou, Olga; Grunewald, Stephanie; Kingma, Sandra D. K.; Knerr, Ina; Leão-Teles, Elisa; Möslinger, Dorothea; Murphy, Elaine; Őunap, Katrin; Pané, Adriana; Paci, Sabrina; Parini, Rossella; Rivera, Isabel A.; Scholl-Bürgi, Sabine; Schwartz, Ida V. D.; Sdogou, Triantafyllia; Shakerdi, Loai A.; Skouma, Anastasia; Stepien, Karolina M.; Treacy, Eileen P.; Waisbren, Susan; Berry, Gerard T.; Rubio-Gozalbo, M. Estela	
Troponin u urinu kao marker srčanoga oštećenja u djece	Bakoš, Matija	
Klinička, neuroslikovna i genetička obilježja pacijenata sa sindromom Leigh	Miljanić, Klara	
Metaboličke miopatije	Bukovac, Antonia	
Fenilketonurija i trudnoća	Benčić, Jelena	
Poremećaj metabolizma purina i pirimidina u djece	Bobek, Klara	
Genotype-phenotype correlation in contactin-associated protein-like 2 (CNTNAP-2) developmental disorder	D'Onofrio, Gianluca; Accogli, Andrea; Severino, Mariasavina; Caliskan, Haluk; Kokotović, Tomislav; Blažeković, Antonela; Gotovac Jerčić, Kristina; Markovic, Silvana; Žigman, Tamara; Goran, Krnjak; Barišić, Nina; Duranovic, Vlasta; Ban, Ana; Borovečki, Fran; Petković Ramadža, Danijela; Barić, Ivo; Fazeli, Walid; Herkenrath, Peter; Marini, Carla; Vittorini, Roberta; Gowda, Vykuntaraju; Bouman, Arjan; Rocca, Clarissa; Alkhawaja, Issam Azmi; Murtaza, Bibi Nazia; Rehman, Malik Mujaddad Ur; Al Alam, Chadi; Nader, Gisele; Mancardi, Maria Margherita; Giacomini, Thea; Srivastava, Siddharth; Alvi, Javeria Raza; Tomoum, Hoda; Matricardi, Sara; Iacomino, Michele; Riva, Antonella; Scala, Marcello; Madia, Francesca; Pistorio, Angela; Salpietro, Vincenzo; Minetti, Carlo; Rivièrè, Jean-Baptiste; Srouf, Myriam; Efthymiou, Stephanie; Maroofian, Reza; Houlden, Henry; Vernes, Sonja Catherine; Zara, Federico; Striano, Pasquale; Nagy, Vanja	
Iskustva s galaktozemijom u Hrvatskoj	Šmaguc, Ana; Ramadža, Danijela Petković; Sarnavka, Vladimir; Krželj, Vjekoslav; Lozić, Bernarda; Pušeljić, Silvija; Rahelić, Valentina; Mesarić, Nikola; Grubić, Marina; Bogdanić, Ana; Špehar Uroić, Anita; Žigman, Tamara; Grizelj, Ruža; Vuković, Jurica; Mardešić, Duško; Szatmari, Ildiko; Rivera, Isabel; Fumić, Ksenija; Barić, Ivo	

Impact of the SARS-CoV-2 pandemic on the health of individuals with intoxication-type metabolic diseases- Data from the E-IMD consortium	Mütze, Ulrike; Gleich, Florian; Barić, Ivo; Baumgartner, Mathias; Burlina, Alberto; Chapman, Kimberly A.; Chien, Yin-Hsiu; Cortès-Saladelafont, Elisenda; De Laet, Corinne; Dobbelaere, Dries; Eysken, Francois; Gautschi, Matthias; Santer, Rene; Häberle, Johannes; Joaquín, Clara; Karall, Daniela; Lindner, Martin; Lund, Allan M.; Mühlhausen, Chris; Murphy, Elaine; Roland, Dominique; Ruiz Gomez, Angeles; Skouma, Anastasia; Grünert, Sarah C.; Wagenmakers, Margreet; Garbade, Sven F.; Kölker, Stefan; Boy, Nikolas	
Prirođeni poremećaji glikozilacije	Pintarić, Martina	
Hipofosfatemični rahitis u djece	Ivanković, Katarina	
Case Report: Advanced Skeletal Muscle Imaging in S-Adenosylhomocysteine Hydrolase Deficiency and Further Insight Into Muscle Pathology	Petković Ramadža, Danijela; Kuhtić, Ivana; Žarković, Kamelija; Lochmüller, Hanns; Čavka, Mislav; Kovač, Ida; Barić, Ivo; Prutki, Maja	
Genetics of Pediatric Epilepsy : Next-Generation Sequencing in Clinical Practice	Blazekovic, Antonela; Gotovac Jercic, Kristina; Meglaj, Sarah; Duranovic, Vlasta; Prpic, Igor; Lozic, Bernarda; Malenica, Masa; Markovic, Silvana; Lujic, Lucija; Gadze, Zeljka Petelin; Juraski, Romana Gjergja; Barisic, Nina; Baric, Ivo; Borovecki, Fran	
NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency	Lenz, Dominic; Pahl, Jens; Hauck, Fabian; Alameer, Seham; Balasubramanian, Meena; Barić, Ivo; Boy, Nikolas; Church, Joseph A.; Crushell, Ellen; Dick, Anke; Distelmaier, Felix; Gujar, Jidnyasa; Indolfi, Giuseppe; Lurz, Eberhard; Peters, Bianca; Schwerd, Tobias; Serranti, Daniele; Kölker, Stefan; Klein, Christoph; Hoffmann, Georg F.; Prokisch, Holger; Greil, Johann; Cerwenka, Adelheid; Giese, Thomas; Staufner, Christian	
Kvaliteta života pacijenata s fenilketonurijom u Hrvatskoj	Alaber, Maja	
Porodična hiperkolesterolemija u djece	Jelovčić, Fabijan	
Inborn Errors of Metabolism Associated With Autism Spectrum Disorders: Approaches to Intervention	Žigman, Tamara; Petković Ramadža, Danijela; Šimić, Goran; Barić, Ivo	
Dijagnostički izazovi, klinička slika i tijek klasične galaktozemije	Šmaguc, Ana	
Hipofosfatazija u djece	Smajo, Ana	

Defining clinical subgroups and genotype-phenotype correlations in NBAS-associated disease across 110 patients		Staufner, Christian; Peters, Bianca; Wagner, Matias; Alameer, Seham; Barić, Ivo; Broué, Pierre; Bulut, Derya; Church, Joseph A.; Crushell, Ellen; Dalgıç, Buket; Das, Anibh M.; Dick, Anke; Dikow, Nicola; Dionisi-Vici, Carlo; Distelmaier, Felix; Bozbulut, Neslihan Ekşi; Feillet, François; Gonzales, Emmanuel; Hadzic, Nedim; Hauck, Fabian; Hegarty, Robert; Hempel, Maja; Herget, Theresia; Klein, Christoph; Konstantopoulou, Vassiliki; Kopajtich, Robert; Kuster, Alice; Laass, Martin W.; Lainka, Elke; Larson-Nath, Catherine; Leibner, Alexander; Lurz, Eberhard; Mayr, Johannes A.; McKiernan, Patrick; Mention, Karine; Moog, Ute; Mungan, Neslihan Onenli; Riedhammer, Korbinian M.; Santer, René; Palafoll, Irene Valenzuela; Vockley, Jerry; Westphal, Dominik S.; Wiedemann, Arnaud; Wortmann, Saskia B.; Diwan, Gaurav D.; Russell, Robert B.; Prokisch, Holger; Garbade, Sven F.; Kölker, Stefan; Hoffmann, Georg F.; Lenz, Dominic	
Omjer S-adenozilmetionina i S-adenozilhomocisteina i polimorfizmi gena za S-adenozilhomocistein hidrolazu u novorođenčadi s prirođenim srčanim greškama i njihovih majki		Ninković, Dorotea	
Klinička i biokemijska obilježja djece s poremećajima beta-oksidacije masnih kiselina		Mlinarić, Stella	
Loše regulirana fenilketonurija s prikazom bolesnika		Marinović, Lucija	
Metilmalonska acidurija		Šikić, Katarina	
Progresivna osificirajuća fibrodisplazija		Gatin, Neva	
Nedostatna aktivnost S-adenozilhomocistein hidrolaze i omjer S-adenozilmetionina i S-adenozilhomocisteina u osoba s trajno povišenom aktivnošću kreatin kinaze		Petković Ramadža, Danijela	
Gaucher disease in children		Shoham, Ori Zion	
Plućna hipertenzija u djece s prirođenim srčanim grješkama		Vukšević, Tonka	

A novel PGAP3 mutation in a Croatian boy with brachytelephalangy and a thin corpus callosum		Sakaguchi, Tomohiro; Žigman, Tamara; Petković Ramadža, Danijela; Omerza, Lana; Pušeljić, Silvija; Ereš Hrvaćanin, Zrinka; Miyake, Noriko; Matsumoto, Naomichi; Barić, Ivo	
Attention deficit/hyperactivity disorder as an associated feature in OCTN2 deficiency with novel deletion (p.T440-Y449)		Lamhonwah, Anne-Marie; Barić, Ivo; Lamhonwah, Jessica; Grubić, Marina; Tein, Ingrid	
The genotypic and phenotypic spectrum of MTO1 deficiency		O'Byrne, James J.; Tarailo-Graovac, Maja; Ghani, Aisha; Champion, Michael; Deshpande, Charu; Dursun, Ali; Ozgul, Riza K.; Freisinger, Peter; Garber, Ian; Haack, Tobias B.; Horvath, Rita; Barić, Ivo; Husain, Ralf A.; Kluijtmans, Leo A.J.; Kotzaeridou, Urania; Morris, Andrew A.; Ross, Colin J.; Santra, Saikat; Smeitink, Jan; Tarnopolsky, Mark; Wortmann, Saskia B.; Mayr, Johannes A.; Brunner-Krainz, Michaela; Prokisch, Holger; Wasserman, Wyeth W.; Wevers, Ron A.; Engelke, Udo F.; Rodenburg, Richard J.; Ting, Teck Wah; McFarland, Robert; Taylor, Robert W.; Salvarinova, Ramona; van Karnebeek, Clara D.M.	
A Novel PGAP3 Mutation in a Croatian boy with Brachytelephalangy and a Thin Corpus Callosum		Sakaguchi, Tomohiro; Žigman, Tamara; Petković Ramadža, Danijela; Omerza, Lana; Pušeljić, Silvija; Ereš Hrvaćanin, Zrinka; Miyake, Noriko; Matsumoto, Naomichi; Barić, Ivo	
Consensus recommendations for the diagnosis, treatment and follow-up of inherited methylation disorders		Barić, Ivo; Staufner, Christian; Augoustides-Savvopoulou, Persephone; Chien, Yin-Hsiu; Dobbelaere, Dries; Grünert, Sarah C.; Opladen, Thomas; Petković Ramadža, Danijela; Rakić, Bojana; Wedell, Anna; Blom, Henk J.	
Etiološki pristup hipoglikemiji u djece		Romić, Tena Nicole	
Molarno incizivna hipomineralizacija u djece s intelektualnim teškoćama		Modrić, Vesna-Erika	
Abnormal hypermethylation at imprinting control regions in patients with S-adenosylhomocysteine hydrolase (AHCY) deficiency		Motzek, Antje; Knežević, Jelena; Switzeny, Olivier J.; Cooper, Alexis; Barić, Ivo; Belužić, Robert; Strauss, Kevin A.; Puffenberger, Erik G.; Mudd, S. Harvey; Vugrek, Oliver; Zechner, Ulrich	
Review and evaluation of the methodological quality of the existing guidelines and recommendations for inherited neurometabolic disorders		Cassis, Linda; Cortès-SaladelaFont, Elisenda; Molero-Luis, Marta; Yubero, Delia; González, Maria Julieta; Herrero, Aida Ormazabal; Fons, Carme; Jou, Cristina; Sierra, Cristina; Castejon Ponce, Esperanza; Ramos, Federico; Armstrong, Judith; O'Callaghan, M. Mar; Casado, Mercedes; Montero, Raquel; Olivas, Silvia Maria Meavilla; Artuch, Rafael; Barić, Ivo; Bartoloni, Franco; Bellettato, Cinzia Maria; Bonifazi, Fedele; Ceci, Adriana; Cvitanović-Šojat, Ljerka; Dali, Christine I; D'Avanzo, Francesca; Fumić, Ksenija; Giannuzzi, Viviana; Lampe, Christina; Scarpa, Maurizio; Garcia-Cazorla, Ángels	

Klinička raznolikost bolesti uzrokovane nasljednim mutacijama gena za prijenosnik tiamina - 2 (ThTr2)	Debeljak, Lucija	
Prenatalna kardiologija	Levicki, Rea	
Biallelic mutations in NBAS cause recurrent acute liver failure with onset in infancy	Haack, Tobias B.; Staufner, Christian; Köpke, Marlies G.; Straub, Beate K.; Kölker, Stefan; Thiel, Christian; Freisinger, Peter; Barić, Ivo; McKiernan, Patrick J.; Dikow, Nicola; Harting, Inga; Beisse, Flemming; Burgard, Peter; Kotzaeridou, Urania; Kühr, Joachim; Himbert, Urban; Taylor, Robert W.; Distelmaier, Felix; Vockley, Jerry; Ghaloul-Gonzalez, Lina; Zschocke, Johannes; Kremer, Laura S.; Graf, Elisabeth; Schwarzmayr, Thomas; Bader, Daniel M.; Gagneur, Julien; Wieland, Thomas; Terrile, Caterina; Strom, Tim M.; Meitinger, Thomas; Hoffmann, Georg F.; Prokisch, Holger	
Manjak lizosomske kisele lipaze u djece: vlastita iskustva i nova mogućnost enzimskoga nadomjesnog liječenja	Petković Ramadža, Danijela; Ćuk, Mario; Zibar, Karin; Barić, Marina; Sarnavka, Vladimir; Bilić, Karmen; Fumić, Ksenija; Vuković, Jurica; Pušeljić, Silvija; Ćorić, Marijana; Štern Padovan, Ranka; Kralik, Marko; Barić, Ivo	
Neonatal liver failure	Ćaćić, Ivana Marija	
Oblici kardiomiopatija u djece	Kresina, Barbara	
Utjecaj nedostatne aktivnosti S-adenozilhomocistein hidrolaze na metilaciju proteina	Ćuk, Mario	
Kongenitalni hiperinzulinizam - novosti o nastanku, dijagnosticiranju i liječenju bolesti	Martinac, Iva; Bogović, Marko; Batinica, Stipe; Sarnavka, Vladimir; Huljev Frković, Sanda; Matić, Toni; Jakić-Razumović, Jasminka; Rubin, Otmar; Luetić, Tomislav; Kušec, Vesna; Petković Ramadža, Danijela; Begović, Davor; Benjak, Vesna; Dasović-Buljević, Andrea; Antabak, Anko; Ćavar, Stanko; Kukin, Dijana; Sršen-Medančić, Suzana; Barić, Ivo	
Spontaneous perforation of the small intestine, a novel manifestation of classical homocystinuria in an adult with new cystathionine beta-synthetase gene mutations	Muačević-Katanec, Diana; Kekez, Tihomir; Fumić, Ksenija; Barić, Ivo; Merkler, Marijan; Jakić-Razumović, Jasminka; Krznarić, Željko; Zadro, Renata; Katanec, Davor; Reiner, Željko	
Manjak vitamina b12 u djece – podcijenjena opasnost u svjetlu novih spoznaja	Juras, Karin; Fumić, Ksenija; Ižaković, Senka; Pušeljić, Silvija; Čulić, Vida; Galić, Slobodan; Dasović Buljević, Andrea; Benjak, Vesna; Čolić, Ana; Huljev Frković, Sanda; Maradin, Miljenka; Sarnavka, Vladimir; Bilić, Karmen; Baumgartner, Matthias; Barić, Ivo	

Pallister Killian syndrome: unusual significant postnatal overgrowth in a girl with otherwise typical presentation		Huljev Frković, Sanda; Tonković Đurišević, Ivana; Lasan Trčić, Ružica; Sarnavka, Vladimir; Crkvenac Gornik, Kristina; Mužinić, Dubravka; Letica, Ljiljana; Barić, Ivo; Begović, Davor	
Hypophosphatasia: phenotypic variability and possible Croatian origin of the c.1402g>A mutation of TNSALP gene		Petković Ramadža, Danijela; Stipoljev, Feodora; Sarnavka, Vladimir; Begović, Davor; Potočki, Kristina; Fumić, Ksenija; Mornet, Etienne; Barić, Ivo	
Fumaric aciduria: mild phenotype in a 8-year-old girl with novel mutations		Maradin, Miljenka; Fumić, Ksenija; Hansikova, H.; Tesarova, M.; Wenchich, L.; Dorner, Sanja; Sarnavka, Vladimir; Zeman, J.; Barić, Ivo	