

Vrijeme izvoza: 25.04.2024. 05:32:14

Repozitorij: repozitorij.kbco.hr

Ukupan broj zapisa na URL-u: 49

Broj izvezenih zapisa: 49

Naslov	URL	Autori	Naslov izvornika
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, Angels	

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; Schwarzmayer, 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	