Magdalena Bartnik

Lista publikacji
z dnia 31 października 2012

Publikacje w czasopismach

1. Bartnik M, Szczepanik E, Derwińska K, Wiśniowiecka-Kowalnik B, Gambin T, Sykulski M, Ziemkiewicz K, Kędzior M, Gos M, Hoffman-Zacharska D, Mazurczak T, Jeziorek A, Antczak-Marach D, Rudzka-Dybała M, Mazurkiewicz H, Goszczańska-Ciuchta A, Zalewska-Miszkurka Z, Terczyńska I, Sobierajewicz M, Shaw CA, Gambin A, Mierzewska H, Mazurczak T, Obersztyn E, Bocian E, Stankiewicz P., 2012, ***Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders***, *American Journal of Medical Genetics Part B Neuropsychiatric Genetics 159B:760-771*

2. Derwińska K, Bartnik M, Wiśniowiecka-Kowalnik B, Jagła M, Rudziński A, Pietrzyk JJ, Kawalec W, Ziółkowska L, Kutkowska-Kaźmierczak A, Gambin T, Sykulski M, Shaw CA, Gambin A, Mazurczak T, Obersztyn E, Bocian E, Stankiewicz P., 2012, ***Assessment of the role of copy-number variants in 150 patients with congenital heart defects***, *Medycyna Wieku Rozwojowego XVI (3):175-182*

3. Wiśniowiecka-Kowalnik B, Kastory-Bronowska M, Bartnik M, Derwińska K, Dymczak-Domini W, Szumbarska D, Ziemka E, Szczałuba K, Sykulski M, Gambin T, Gambin A, Shaw CA, Mazurczak T, Obersztyn E, Bocian E, Stankiewicz P., 2012, ***Application of custom-designed oligonucleotide array CGH in 145 patients with autistic spectrum disorders***, *European Journal of Human Genetics (publikacja w wersji elektronicznej doi:10.1038/ejhg.2012.219)*

4. Bilir B, Yapici Z, Yalcinkaya C, Baris I, Carvalho C, Bartnik M, Ozes B, Eraksoy M, Lupski J, Battaloglu E., 2012, ***High frequency of GJA12/GJC2 mutations in Turkish patients with Pelizaeus-Merzbacher disease***, *Clinical Genetics (publikacja w wersji elektronicznej doi: 10.1111/j.1399-0004.2012.01846.x.)*

5. Carvalho CMB, Bartnik M, Pehlivan D, Fang P, Shen J, Lupski JR., 2012, ***Evidence for disease penetrance relating to CNV size: Pelizaeus–Merzbacher disease and manifesting carriers with a familial 11 Mb duplication at Xq22***, *Clinical Genetics 81:532-534*

6. Bartnik M., Derwinska K., Gos M., Obersztyn E., Kołodziejska K.E., Erez A., Szpecht–Potocka A., Fang P., Terczynska I., Mierzewska H., Lohr N.J., Bellus G.A., Reimschisel T., Bocian E., Mazurczak T., Cheung SW., Stankiewicz P., 2011, ***Early-onset seizures due to mosaic exonic deletions of CDKL5 in a male and two females***, *Genetics in Medicine 13(5): 447-452*

7. Bartnik M., Chun-Hui Tsai A., Xia Z., Cheung S.W., Stankiewicz P., 2011, ***Disruption of the SCN2A and SCN3A genes in a patient with mental retardation, neurobehavioral and psychiatric abnormalities, and a history of infantile seizures***, *Clinical Genetics 80: 191-195*

8. Boone P.M., Bacino C.A., Shaw C.A., Eng P.A., Hixson P.M., Pursley A.N., Kang S.H., Yang Y., Wiszniewska J., Nowakowska B.A., Del Gaudio D., Xia Z., Simpson-Patel G., Immken L.L., Gibson J.B., Tsai A.C., Bowers J.A., Reimschisel T.E., Schaaf C.P., Potocki L., Scaglia F., Gambin T., Sykulski M., Bartnik M., Derwinska K., Wisniowiecka-Kowalnik B., Lalani S.R., Probst F.J., Bi W., Beaudet A.L., Patel A., Lupski J.R., Cheung S.W., Stankiewicz P., 2010, ***Detection of clinically relevant exonic copy-number changes by array CGH***, *Human Mutation 31(12): 1326–1342*

9. Ramocki M.B., Bartnik M., Szafranski P., Kołodziejska K.E., Xia Z., Bravo J., Miller G.S., Rodriguez D.L., Williams C.A., Bader P.I., Szczepanik E., Mazurczak T., Antczak-Marach D., Coldwell J.G., Akman C.I., McAlmon K., Cohen M.P., McGrath J., Roeder E., Clark G.D., Mueller J., Kang S.H.L., Bacino C.A., Patel A., Bocian E., Shaw C., Cheung S.W., Mazurczak T., Stankiewicz P., 2010, ***Recurrent distal 7q11.23 deletion***

including HIP1 and YWHAG identified in patients with epilepsy, learning difficulties, intellectual disabilities, and neurobehavioral abnormalities, *American Journal of Human Genetics 87: 857–865*