

# Author Index Volume 1 (2012)

The issue number is given in front of the pagination

- Abdallah Bouhjar, I.B., A. Gmidène, N. Soyah, H. Hanene, S. Mougou, H. Elghezal and A. Saad, Trisomy and tetrasomy 15q11-q13 diagnosed by molecular cytogenetic analysis in two patients with mental retardation (1) 63–68
- Abdallah Bouhjar, I.B., A. Gmidène, S. Mougou-Zrelli, H. Hannachi, N. Soyah, N. Gadour, I. Harrabi, H. Elghezal and A. Saad, Cytogenetic analysis in a large series of children with non-syndromic mental retardation (3) 175–180
- Akbaş, H., see Balkan, M. (4) 243–246
- Akrami, S.M., Genetics of consanguineous marriage: impact and importance of counseling (4) 217–220
- Albano, A., see Wang, K. (2) 85–98
- Alp, M.N., see Balkan, M. (4) 239–242
- Alp, M.N., see Balkan, M. (4) 243–246
- An, K., see Chen, X. (1) 39–45
- Andrade, E. and C. Williams, The importance of developing novel diagnostic tools for congenital metabolic disorders (3) 149–151
- Anguiano, A., see Hantash, F.M. (2) 115–124
- Ashavaid, T.F., see Deepak, R.R. (4) 221–227
- Avard, D., see Greenberg, C. (1) 7–11
- Babcock, M., see Samanich, J. (1) 47–53
- Balasubramanian, M., K. Smith, S. Williams, P.D. Griffiths, M.J. Parker and S.R. Mordekar, Tigroid pattern of cerebral white matter involvement in chromosome 6p25 deletion syndrome with concomitant 5p15 duplication (4) 247–252
- Balkan, M., M. Fidanboy, C. Özmen, M.N. Özbek, S. Otçu, E. Kapı and T. Budak, Cytogenetic and clinical features of a 13 year old male with trisomy 8 (3) 205–208
- Balkan, M., M. Fidanboy, H. İsi, H. Akbaş, S. Kalkanlı, M.N. Alp and T. Budak, A case of complete tetraploidy in amniocentesis with normal karyotype in subsequent cordocentesis (4) 243–246
- Balkan, M., M. Fidanboy, M.N. Özbek, M.N. Alp and T. Budak, Different chromosome Y abnormalities in a case with short stature (4) 239–242
- Bender, A., see Wang, K. (2) 85–98
- Böckenhauer, D., A. Bökenkamp, M. Nuutinen, R. Unwin, W. van't Hoff, T. Sirimanna, K. Vrljicak and M. Ludwig, Novel *OCRL* mutations in patients with Dent-2 disease (1) 15–23
- Bökenkamp, A., see Böckenhauer, D. (1) 15–23
- Bowers, M. and G. Gold-von Simson, Moebius syndrome with baroreflex failure in an adolescent female (3) 199–203
- Boyar, F.Z., see Hantash, F.M. (2) 115–124
- Bradfield, J.P., see Wang, K. (2) 85–98
- Budak, T., see Balkan, M. (3) 205–208
- Budak, T., see Balkan, M. (4) 239–242
- Budak, T., see Balkan, M. (4) 243–246
- Chandler-Laney, P.C., see De Luca, M. (4) 229–234
- Chen, G., see Chen, X. (1) 39–45
- Chen, X., J. Tang, Y. Liu, M. Luan, K. An, Y. Zhang, F. Li, P. Zhou, W. Liu, J. Liu and G. Chen, Lack of association between *NCAM1* and early onset schizophrenia in a family based study in Shandong peninsula of China (1) 39–45
- Chiavacci, R.M., see Wang, K. (2) 85–98
- Citrigno, L., see Patitucci, A. (2) 99–102
- Cohen, L., J. Samanich, Q. Pan, L. Mehta and R. Marion, 17q12 Deletion in a patient with Williams syndrome: Case report and review of the literature (2) 135–141
- Conforti, F.L., see Patitucci, A. (2) 99–102
- Cornforth, C.M., see Morgan, A.R. (2) 103–113

- Crespo, F., H. Pinar and S. Kostadinov, Cases of limb-body wall complex: Early amnion rupture, vascular disruption, or abnormal splitting of the embryo? (4) 235–238
- Dalal, S., see Sinha, R. (3) 195–197
- De Luca, M., P.C. Chandler-Laney, H. Wiener and J.R. Fernandez, Common variants in the *LAMA5* gene associate with fasting plasma glucose and serum triglyceride levels in a cohort of pre- and early pubertal children (4) 229–234
- de Paula Queiroz, R.G., see Mateo, E.C. (3) 181–187
- Deepak, R.R. and T.F. Ashavaid, Screening of the CFTR gene in Indian patients (4) 221–227
- Denecke, B., see Eggermann, T. (2) 143–147
- Dinour, D., see Levin-Iaina, N. (1) 3–5
- Dorobisz, U., see Smigiel, R. (1) 55–58
- Eggermann, T., S. Spengler, U. Gamerding, B. Denecke, S. Grimm, M. Grimm, R. Schubert and G. Schwanitz, Duplication 3q13.11q23: Longitudinal study in a patient over a period of more than 7 years and refinements of the breakpoints (2) 143–147
- El-Ruby, M.O., see Ismail, S. (3) 189–194
- Elghezal, H., see Abdallah Bouhjar, I.B. (1) 63–68
- Elghezal, H., see Abdallah Bouhjar, I.B. (3) 175–180
- Fabregat, M., see Mimbacas, A. (2) 131–134
- Faith, M.S., Electronic medical records, genetics, and childhood obesity: A new direction for scientific discovery? (2) 69–70
- Farias, J., see Mimbacas, A. (2) 131–134
- Ferguson, L.R., see Morgan, A.R. (2) 103–113
- Fernández, M., see Mimbacas, A. (2) 131–134
- Fernandez, J.R., see De Luca, M. (4) 229–234
- Fidanboy, M., see Balkan, M. (3) 205–208
- Fidanboy, M., see Balkan, M. (4) 239–242
- Fidanboy, M., see Balkan, M. (4) 243–246
- Flore, L.A., E. Leon, T.A. Maher and J.M. Milunsky, *RASA1* analysis guides management in a family with capillary malformation-arteriovenous malformation (2) 125–129
- Frackelton, E.C., see Wang, K. (2) 85–98
- Gabbett, M.T., The oculoauriculovertebral spectrum: Refining the estimate of birth prevalence (2) 71–77
- Gabriele, A.L., see Patitucci, A. (2) 99–102
- Gadour, N., see Abdallah Bouhjar, I.B. (3) 175–180
- Gamerding, U., see Eggermann, T. (2) 143–147
- Garris, M., see Wang, K. (2) 85–98
- Girisha, K.M., see Kodandapani, S. (1) 59–61
- Glessner, J.T., see Wang, K. (2) 85–98
- Gmidène, A., see Abdallah Bouhjar, I.B. (1) 63–68
- Gmidène, A., see Abdallah Bouhjar, I.B. (3) 175–180
- Gold-von Simson, G., see Bowers, M. (3) 199–203
- Golebiowski, W., see Smigiel, R. (1) 55–58
- Grant, S.F.A., see Wang, K. (2) 85–98
- Greenberg, C., K. McClellan and D. Avard, Beyond dissemination: A knowledge translation model to drive change in pediatric genetics (1) 7–11
- Griffiths, P.D., see Balasubramanian, M. (4) 247–252
- Grimm, M., see Eggermann, T. (2) 143–147
- Grimm, S., see Eggermann, T. (2) 143–147
- Grundmeier, R., see Wang, K. (2) 85–98
- Guo, Y., see Wang, K. (2) 85–98
- Hakonarson, H., see Wang, K. (2) 85–98
- Hanene, H., see Abdallah Bouhjar, I.B. (1) 63–68
- Hannachi, H., see Abdallah Bouhjar, I.B. (3) 175–180
- Hantash, F.M., B.T. Wang, R. Owen, L.P. Ross, L.W. Mahon, F.Z. Boyar, A. Anguiano and C.M. Strom, Inherited and *de novo* 22q11.2 distal duplications in two patients with autistic features, speech delay and no dysmorphology (2) 115–124
- Harrabi, I., see Abdallah Bouhjar, I.B. (3) 175–180
- Helmy, N.A., see Ismail, S. (3) 189–194
- Horth, L., M.W. Stacey, V.K. Proud, K. Segna, C. Rutherford, D. Nuss and R.E. Kelly, Advancing our understanding of the inheritance and transmission of pectus excavatum (3) 161–173
- Hou, C., see Wang, K. (2) 85–98
- İsi, H., see Balkan, M. (4) 243–246
- Ismail, S., N.A. Helmy, W.M. Mahmoud and M.O. El-Ruby, Phenotypic characterization of rare interstitial deletion of chromosome 4 (3) 189–194
- Janik, D.K., see Lindau-Shepard, B. (3) 153–160
- Javiel, G., see Mimbacas, A. (2) 131–134
- John, B.M., see Sinha, R. (3) 195–197
- Kalkanli, S., see Balkan, M. (4) 243–246
- Kapı, E., see Balkan, M. (3) 205–208
- Keating, B., see Wang, K. (2) 85–98
- Kelly, R.E., see Horth, L. (3) 161–173
- Kim, C.E., see Wang, K. (2) 85–98

- Kobayashi, R., K. Matsune and H. Ohashi, Fused teeth, macrodontia and increased caries are characteristic features of neurofibromatosis type 1 patients with *NFI* gene microdeletion (1) 25–31
- Kodandapani, S., J. Shetty, P. Kumar and K.M. Girisha, Umbilical cyst due to patent urachus in a fetus with complete urorectal septum malformation sequence (1) 59–61
- Kostadinov, S., see Crespo, F. (4) 235–238
- Kumar, P., see Kodandapani, S. (1) 59–61
- Lam, W.-J., see Morgan, A.R. (2) 103–113
- Lebioda, A., see Smigiel, R. (1) 55–58
- Leon, E., see Flore, L.A. (2) 125–129
- Levin-Iaina, N. and D. Dinour, Renal disease with *OCRL1* mutations: Dent-2 or Lowe syndrome? (1) 3–5
- Li, F., see Chen, X. (1) 39–45
- Lindau-Shepard, B., D.K. Janik and K.A. Pass, A microsphere-based assay for mutation analysis of the biotinidase gene using dried blood spots (3) 153–160
- Liu, J., see Chen, X. (1) 39–45
- Liu, W., see Chen, X. (1) 39–45
- Liu, Y., see Chen, X. (1) 39–45
- Luan, M., see Chen, X. (1) 39–45
- Ludwig, M. and H. Reutter, Genome-wide array data and next generation sequencing unravel the etiology of urogenital malformations (4) 209–216
- Ludwig, M., see Böckenhauer, D. (1) 15–23
- Magariello, A., see Patitucci, A. (2) 99–102
- Maher, T.A., see Flore, L.A. (2) 125–129
- Mahmoud, W.M., see Ismail, S. (3) 189–194
- Mahon, L.W., see Hantash, F.M. (2) 115–124
- Marion, R., see Cohen, L. (2) 135–141
- Mateo, E.C., F.J.N. Motta, R.G. de Paula Queiroz, C.A. Scrideli and L.G. Tone, Protein expression of matrix metalloproteinase (MMP-1, -2, -3, -9 and -14) in Ewing family tumors and medulloblastomas of pediatric patients (3) 181–187
- Matsune, K., see Kobayashi, R. (1) 25–31
- Mazzei, R., see Patitucci, A. (2) 99–102
- McClellan, K., see Greenberg, C. (1) 7–11
- Mehta, L., see Cohen, L. (2) 135–141
- Mentch, F.D., see Wang, K. (2) 85–98
- Milunsky, J.M., see Flore, L.A. (2) 125–129
- Mimbacas, A., G. Vitarella, J. Souto, A.L. Reyes, J. Farias, M. Fernández, M. Fabregat and G. Javiel, The phenotype masks the genotype: A possible new expression of diabetes (2) 131–134
- Misiak, B., see Smigiel, R. (1) 55–58
- Mitchell, E.A., see Morgan, A.R. (2) 103–113
- Montagna, C., see Samanich, J. (1) 47–53
- Mordekar, S.R., see Balasubramanian, M. (4) 247–252
- Morgan, A.R., J.M.D. Thompson, K.E. Waldie, C.M. Cornforth, D. Turic, E.J.S. Sonuga-Barke, W.-J. Lam, L.R. Ferguson and E.A. Mitchell, Initial evidence that polymorphisms in neurotransmitter-regulating genes contribute to being born small for gestational age (2) 103–113
- Morrow, B.E., see Samanich, J. (1) 47–53
- Motta, F.J.N., see Mateo, E.C. (3) 181–187
- Mougou, S., see Abdallah Bouhjar, I.B. (1) 63–68
- Mougou-Zrelli, S., see Abdallah Bouhjar, I.B. (3) 175–180
- Moy, A., see Wang, K. (2) 85–98
- Muglia, M., see Patitucci, A. (2) 99–102
- Negi, V., see Sinha, R. (3) 195–197
- Nuss, D., see Horth, L. (3) 161–173
- Nuutinen, M., see Böckenhauer, D. (1) 15–23
- Özbek, M.N., see Balkan, M. (3) 205–208
- Özbek, M.N., see Balkan, M. (4) 239–242
- Özmen, C., see Balkan, M. (3) 205–208
- Ohashi, H., see Kobayashi, R. (1) 25–31
- Otçu, S., see Balkan, M. (3) 205–208
- Otieno, G., see Wang, K. (2) 85–98
- Owen, R., see Hantash, F.M. (2) 115–124
- Pan, Q., see Cohen, L. (2) 135–141
- Parker, M.J., see Balasubramanian, M. (4) 247–252
- Pass, K.A., see Lindau-Shepard, B. (3) 153–160
- Patitucci, A., A. Magariello, C. Ungaro, M. Muglia, F.L. Conforti, A.L. Gabriele, L. Citrigno, W. Sproviero and R. Mazzei, *SMN1* gene copy number analyses for SMA healthy carriers in Italian population (2) 99–102
- Patkowski, D., see Smigiel, R. (1) 55–58
- Pinar, H., see Crespo, F. (4) 235–238
- Proud, V.K., see Horth, L. (3) 161–173
- Qiu, H., see Wang, K. (2) 85–98
- Raju, U., see Sinha, R. (3) 195–197
- Reutter, H., see Ludwig, M. (4) 209–216
- Reyes, A.L., see Mimbacas, A. (2) 131–134

- Ross, L.P., see Hantash, F.M. (2) 115–124  
 Rutherford, C., see Horth, L. (3) 161–173
- Saad, A., see Abdallah Bouhjar, I.B. (1) 63–68  
 Saad, A., see Abdallah Bouhjar, I.B. (3) 175–180  
 Samanich, J., C. Montagna, B.E. Morrow and M. Babcock, Interstitial duplication of 22q13.2 in a girl with short stature, impaired speech and language, and dysmorphism (1) 47–53  
 Samanich, J., see Cohen, L. (2) 135–141  
 Schubert, R., see Eggermann, T. (2) 143–147  
 Schwanitz, G., see Eggermann, T. (2) 143–147  
 Scrideli, C.A., see Mateo, E.C. (3) 181–187  
 Segna, K., see Horth, L. (3) 161–173  
 Seidler, K., see Wang, K. (2) 85–98  
 Shastry, B.S., Pharmacogenomics and its importance in pediatric medicine (2) 79–84  
 Shetty, J., see Kodandapani, S. (1) 59–61  
 Shimojima, K. and T. Yamamoto, Growth profiles of 34 patients with Wolf-Hirschhorn syndrome (1) 33–37  
 Sinha, R., S. Dalal, U. Raju, B.M. John and V. Negi, A case of 9p deletion syndrome with Duane retraction syndrome (3) 195–197  
 Sirimanna, T., see Böckenhauer, D. (1) 15–23  
 Slavotinek, A.M., A much needed new journal in the field of Pediatric Genetics (1) 1–2  
 Sleiman, P.A., see Wang, K. (2) 85–98  
 Smigiel, R., B. Misiak, W. Golebiowski, A. Lebioda, U. Dorobisz, M. Zielinska and D. Patkowski, Esophageal atresia and anal atresia in a newborn with heterotaxia combined with other congenital defects (1) 55–58  
 Smith, K., see Balasubramanian, M. (4) 247–252  
 Sonuga-Barke, E.J.S., see Morgan, A.R. (2) 103–113  
 Souto, J., see Mimbacas, A. (2) 131–134  
 Soyah, N., see Abdallah Bouhjar, I.B. (1) 63–68  
 Soyah, N., see Abdallah Bouhjar, I.B. (3) 175–180  
 Spengler, S., see Eggermann, T. (2) 143–147  
 Sproviero, W., see Patitucci, A. (2) 99–102
- Stacey, M.W., see Horth, L. (3) 161–173  
 Strom, C.M., see Hantash, F.M. (2) 115–124
- Tang, J., see Chen, X. (1) 39–45  
 Thomas, K., see Wang, K. (2) 85–98  
 Thompson, J.M.D., see Morgan, A.R. (2) 103–113  
 Tone, L.G., see Mateo, E.C. (3) 181–187  
 Turic, D., see Morgan, A.R. (2) 103–113
- Ungaro, C., see Patitucci, A. (2) 99–102  
 Unwin, R., see Böckenhauer, D. (1) 15–23
- van't Hoff, W., see Böckenhauer, D. (1) 15–23  
 Vitarella, G., see Mimbacas, A. (2) 131–134  
 Vrljicak, K., see Böckenhauer, D. (1) 15–23
- Waldie, K.E., see Morgan, A.R. (2) 103–113  
 Wang, B.T., see Hantash, F.M. (2) 115–124  
 Wang, K., H. Zhang, F.D. Mentch, J.P. Bradfield, J.T. Glessner, H. Qiu, Y. Guo, C. Hou, E.C. Frackelton, K. Thomas, A. Bender, A. Albano, G. Otieno, M. Garris, K. Seidler, A. Moy, C.E. Kim, B. Keating, R.M. Chiavacci, R. Grundmeier, P.A. Sleiman, S.F.A. Grant and H. Hakonarson, Examination of genetic variants influencing lipid traits in pediatric populations (2) 85–98  
 Wiener, H., see De Luca, M. (4) 229–234  
 Williams, C., see Andrade, E. (3) 149–151  
 Williams, S., see Balasubramanian, M. (4) 247–252  
 Wiwanitkit, V., Prenatal testing: A method for early detection of genetic disorders among fetuses in Thailand, a data between the year 1990 and 2010 (1) 13–14
- Yamamoto, T., see Shimojima, K. (1) 33–37
- Zhang, H., see Wang, K. (2) 85–98  
 Zhang, Y., see Chen, X. (1) 39–45  
 Zhou, P., see Chen, X. (1) 39–45  
 Zielinska, M., see Smigiel, R. (1) 55–58