

## Author Index Volume 3 (2014)

The issue number is given in front of the pagination

- Akrami, S.M. and L. Habibi, Retrotransposons and pediatric genetic disorders: Importance and implications (1) 9–16
- Antoniazzi, F., see Corradi, M. (1) 35–39
- Arno, G., see Sureka, D. (3) 157–162
- Arts, H.H., see Mans, D.A. (2) 47–48
- Au, J., L. Berkowitz-Sutherland, A. Schneider, J.B. Schweitzer, D. Hessel and R. Hagerman, A feasibility trial of Cogmed working memory training in fragile X syndrome (3) 147–156
- Avina, D.A.H., see Fierro, J.A.A. (3) 141–145
- Bergman, M.Y. and S. Nallasamy, Pediatric genetic macular and choroidal diseases (4) 243–258
- Berkowitz-Sutherland, L., see Au, J. (3) 147–156
- Bernstein, J.A., see Sureka, D. (3) 157–162
- Bodurtha, J.N. and J. Duis, Signs and symptoms of genetic conditions – A handbook (3) 183–184
- Butler, M.G., see Jerkovich, A.M. (1) 41–44
- Caldwell, K.S., see Rush, E.T. (1) 29–34
- Callewaert, B., see Mehar, V. (3) 163–166
- Chakravarty, A., see De, P. (3) 175–181
- Chakravarty, S., see De, P. (3) 175–181
- Chatterjee, T., see De, P. (3) 175–181
- Corradi, M., E. Monti, G. Venturi, A. Gandini, M. Mottes and F. Antoniazzi, The recurrent causal mutation for osteogenesis imperfecta type V occurs at a highly methylated CpG dinucleotide within the *IFITM5* gene (1) 35–39
- Coucke, P.J., see Mehar, V. (3) 163–166
- Danda, S., see Ekbote, A.V. (3) 167–173
- Daniels, J.M.A., see Paff, T. (2) 115–127
- De Paepe, A., see Mehar, V. (3) 163–166
- de Vries, T.I. and M.M. van Haelst, Ciliary disturbances in syndromal and non-syndromal obesity (2) 79–88
- De, P., T. Chatterjee, S. Chakravarty and A. Chakravarty, Clinical presentation of two  $\beta$ -thalassemic Indian patients with 1p36 deletion syndrome: Case report (3) 175–181
- Duis, J., see Bodurtha, J.N. (3) 183–184
- Dutta, U.R., Precision in chromosome identification with leads in molecular cytogenetics: An illustrated review (1) 1–7
- Ekbote, A.V., M.S. Kamath and S. Danda, MURCS association with situs inversus totalis: Expanding the spectrum or a novel disorder (3) 167–173
- Esposito, P.W., see Rush, E.T. (1) 29–34
- Fecarotta, C.M. and W.W. Huang, Pediatric genetic disease of the cornea (4) 195–207
- Fierro, J.A.A. and D.A.H. Avina, Pitt-Hopkins syndrome: Mental retardation, psychomotor and developmental delays with facial dysmorphism (3) 141–145
- Fraga, V.G. and K.B. Gomes, Adiponectin gene polymorphisms: Association with childhood obesity (1) 17–28
- Gandini, A., see Corradi, M. (1) 35–39
- Giles, R.H., see Klasson, T.D. (2) 129–140
- Gomes, K.B., see Fraga, V.G. (1) 17–28
- Guay-Woodford, L.M., Autosomal recessive polycystic kidney disease: The prototype of the hepato-renal fibrocystic diseases (2) 89–101
- Haarman, E.G., see Paff, T. (2) 115–127
- Habibi, L., see Akrami, S.M. (1) 9–16
- Hagerman, R., see Au, J. (3) 147–156
- Hartill, V.L., see Szymanska, K. (2) 65–78
- Heidary, G., Congenital optic nerve anomalies and hereditary optic neuropathies (4) 271–280
- Hessel, D., see Au, J. (3) 147–156
- Huang, W.W., see Fecarotta, C.M. (4) 195–207
- Ichhpujani, P. and R.B. Singh, Pediatric genetic diseases causing glaucoma (4) 209–218
- Jerkovich, A.M. and M.G. Butler, Further phenotypic expansion of 15q11.2 BP1-BP2 microdeletion (Burnside-Butler) syndrome (1) 41–44
- Johnson, C.A., see Szymanska, K. (2) 65–78

- Kamath, M.S., see Ekbote, A.V. (3) 167–173  
Klasson, T.D. and R.H. Giles, The role of the cilium in hereditary tumor predisposition syndromes (2) 129–140  
Kreikemeier, R.M., see Rush, E.T. (1) 29–34  
Kumar, R., see Mehar, V. (3) 163–166
- Lutz, R.E., see Rush, E.T. (1) 29–34
- Mans, D.A. and H.H. Arts, Medical genetics of ciliopathies (2) 47–48  
Mehar, V., D. Yadav, R. Kumar, S. Yadav, K. Singh, B. Callewaert, S. Pathan, A. De Paepe and P.J. Coucke, Congenital contractural arachnodactyly due to a novel splice site mutation in the *FBN2* gene (3) 163–166  
Monti, E., see Corradi, M. (1) 35–39  
Mottes, M., see Corradi, M. (1) 35–39  
Murphy, D., see Sureka, D. (3) 157–162
- Nallasamy, S., see Bergman, M.Y. (4) 243–258  
Nihalani, B.R., Pediatric genetic disorders of lens (4) 219–227
- Odent, S., see Sureka, D. (3) 157–162
- Paff, T., J.M.A. Daniels, G. Pals and E.G. Haarman, Primary ciliary dyskinesia: From diagnosis to molecular mechanisms (2) 115–127  
Pals, G., see Paff, T. (2) 115–127  
Patel, M. and A. Ramasubramanian, Introduction to pediatric genetic eye diseases (4) 185–193  
Pathan, S., see Mehar, V. (3) 163–166
- Ramasubramanian, A., see Patel, M. (4) 185–193  
Ramasubramanian, A., see Rouhani, B. (4) 259–269
- Rouhani, B. and A. Ramasubramanian, Pediatric genetic ocular tumors (4) 259–269  
Rush, E.T., K.S. Caldwell, R.M. Kreikemeier, R.E. Lutz and P.W. Esposito, Osteogenesis imperfecta caused by *PPIB* mutation with severe phenotype and congenital hearing loss (1) 29–34
- Sadiq, M.A.A. and M. ur Rehman, Genetics of strabismus and lid diseases (4) 281–290  
Say, E.A.T., Genetic pediatric retinal diseases (4) 229–241  
Sayer, J.A., see Srivastava, S. (2) 103–114  
Schmidts, M., Clinical genetics and pathobiology of ciliary chondrodysplasias (2) 49–64  
Schneider, A., see Au, J. (3) 147–156  
Schweitzer, J.B., see Au, J. (3) 147–156  
Singh, K., see Mehar, V. (3) 163–166  
Singh, R.B., see Ichhpujani, P. (4) 209–218  
Srivastava, S. and J.A. Sayer, Nephronophthisis (2) 103–114  
Stheneur, C., see Sureka, D. (3) 157–162  
Sureka, D., C. Stheneur, S. Odent, G. Arno, D. Murphy and J.A. Bernstein, A recurrent fibrillin-1 mutation in severe early onset Marfan syndrome (3) 157–162  
Szymanska, K., V.L. Hartill and C.A. Johnson, Unraveling the genetics of Joubert and Meckel-Gruber syndromes (2) 65–78
- ur Rehman, M., see Sadiq, M.A.A. (4) 281–290
- van Haelst, M.M., see de Vries, T.I. (2) 79–88  
Venturi, G., see Corradi, M. (1) 35–39
- Yadav, D., see Mehar, V. (3) 163–166  
Yadav, S., see Mehar, V. (3) 163–166