

## Contents

---

Preface

---

**77 Paediatric Thyroid Disorders: New Insights**

Polak, M. (Paris); Szinnai, G. (Basel)

---

Original Papers

---

**79 Somatic Mutations Are Not Observed by Exome Sequencing of Lymphocyte DNA from Monozygotic Twins Discordant for Congenital Hypothyroidism due to Thyroid Dysgenesis**

Magne, F.; Serpa, R.; Van Vliet, G.; Samuels, M.E.; Deladoëy, J. (Montreal, Que.)

**86 Multiple Factors Influencing the Incidence of Congenital Hypothyroidism Detected by Neonatal Screening**

Olivieri, A.; Fazzini, C.; Medda, E. (Rome); The Italian Study Group for Congenital Hypothyroidism

**94 The Role of <sup>123</sup>I Imaging in the Evaluation of Infants with Mild Congenital Hypothyroidism**

Graber, E.; Regelman, M.O.; Annunziato, R.; Machac, J.; Rapaport, R. (New York, N.Y.)

**102 Natural History and Management of Congenital Hypothyroidism with in situ Thyroid Gland**

Castanet, M. (Paris/Rouen); Goischke, A.; Léger, J.; Thalassinou, C. (Paris); Rodrigue, D. (Le Kremlin-Bicêtre); Cabrol, S.; Zenaty, D.; al-Harbi, M.; Polak, M.; Czernichow, P. (Paris) on behalf of Fédération Parisienne pour le Dépistage et la Prévention des Handicaps de l'Enfant (FDPHE)

**111 Diagnosis and Management of Congenital Hypothyroidism Associated with Pseudohypoparathyroidism**

Picard, C.; Decrequey, A.; Guenet, D. (Caen); Bursztejn, A.C. (Nancy); Toledano, D.; Richard, N.; Kottler, M.-L. (Caen)

**118 Thyroid Function in Rett Syndrome**

Stagi, S. (Florence); Cavalli, L. (Pisa); Congiu, L.; Scusa, M.F. (Viareggio); Ferlini, A.; Bigoni, S. (Ferrara); Benincasa, A. (Viareggio); Rossi, B. (Pisa); Pini, G. (Viareggio)

---

Review of a Controversial Topic

---

**126 Down Syndrome and Nonautoimmune Hypothyroidisms in Neonates and Infants**

Kariyawasam, D.; Carré, A.; Luton, D.; Polak, M. (Paris)

**132 Severe Unsuspected Maternal Hypothyroidism Discovered after the Diagnosis of Thyrotropin Receptor-Blocking Antibody-Induced Congenital Hypothyroidism in the Neonate: Failure to Recognize and Implications to the Fetus**

Brown, R.S.; Alter, C.A. (Worcester, Mass.); Sadeghi-Nejad, A. (Boston, Mass.)

**136 Foetal Thyroid Dysfunction: Treat the Mother First!**

Khater, C.; Ceccaldi, P.F.; Poujade, O.; Banigé, M.; Ottenwalter, A.; Luton, D. (Clichy)

New Hypothesis

---

**141 Molecular Insights into the Possible Role of Kir4.1 and Kir5.1 in Thyroid Hormone Biosynthesis**

Ramos, H.E. (Paris/Salvador); Dias da Silva, M.R. (Sao Paulo); Carré, A. (Paris); Silva Jr., J.C. (Salvador); Paninka, R.M. (Sao Paulo); Oliveira, T.L. (Salvador); Tron, E. (Paris); Castanet, M. (Paris/Rouen); Polak, M. (Paris)

**148 Author Index/Subject Index**