Paediatric Thyroid Disorders: New Insights

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Many clinical problems in the field of thyroid disease in paediatric patients remain unresolved. Therefore, the 2015 'special topic issue' of *Hormone Research in Paediatrics* focuses on new developments and new knowledge in this field, and some of the papers selected will add new insights while others provide new hypotheses.

Although this special issue focuses on clinical problems, the first article adds an important piece of knowledge on a long-standing question in the context of sporadic rather than inherited congenital hypothyroidism due to thyroid dysgenesis. The fact that monozygotic twins are discordant for thyroid dysgenesis led the authors to hypothesize that rather non-Mendelian than Mendelian mechanisms could be involved in thyroid dysgenesis. They performed whole exome sequencing in lymphocytes of three monozygotic twin pairs discordant for thyroid dysgenesis without evidence of differences in the protein-coding genome using current technology. Although their initial hypothesis of possible early somatic mutations in the affected monozygotic twins was not confirmed, thus presenting 'negative' results, the data deserve special consideration. On the one hand, the authors themselves point to the fact that somatic mutations have not been excluded as the cause of thyroid dysgenesis; on the other hand, they suggest another genetic mechanism:

random monoallelic expression. Thus, the hypothesis of non-Mendelian inheritance remains valid, and the search for other mechanisms ensues. They do not exclude a complex situation in which both Mendelian and non-Mendelian mechanisms are at play.

In the clinical part of this special issue, thyroid disorders associated with well-known genetic syndromes are carefully analyzed and reviewed. First, hypothyroidism is a typical feature of pseudohypoparathyroidism (OMIM 103580), but due to its mild course, it remains to be determined whether compensated hypothyroidism is present at birth or develops later in life, and at what time it should be looked for. A large retrospective study presents clear answers. In a second work, the clinical features of thyroid disorders in the Rett syndrome (OMIM 613454) are outlined. Finally, a detailed update is given on mechanisms of non-autoimmune causes of hypothyroidism in Down's syndrome (OMIM 190685). All three papers provide important information and updates on the current state of knowledge and on future directions of research.

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Maternal thyroid disease can have detrimental effects on fetal thyroid function. Two succinct clinical descriptions provide important insights into possible pitfalls during the clinical follow-up of pregnant women: oligo-symptomatic severe hypothyroidism of the mother detected following neonatal screening of the child, and the pragmatic approach to treat thyroid disease diagnosed during pregnancy in both the fetus and the mother.

For many aspects of congenital hypothyroidism, guidelines have recently been published by the European Society for Paediatric Endocrinology [1]. Nevertheless, novel data concerning screening, diagnosis and management are important for a constant broadening of our knowledge and expertise in the key aspects of the most prevalent neonatal endocrine disorder. Three papers are adding new data concerning factors influencing the detection rate of neonatal screening, the role of scintigraphy in patients suffering from mild congenital hypothyroidism and the natural history of hypothyroidism in patients with thyroid gland 'loco classico'.

In summary, this special topic issue aims to provide new insights into fetal, neonatal and childhood thyroidology in patients suffering from congenital hypothyroidism and thyroid disease associated to genetic syndromes, and in children exposed to maternal thyroid disease. We hope you will enjoy reading.

Disclosure Statement

The authors declare that no conflicts of interest exist.

Reference

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