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Special issue on molecular genetics in endocrinology

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Special issue on molecular genetics in endocrinology

Edição especial em genética molecular em endocrinologia

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Welcome to the 2012 edition of the ABEM "Clinical & Molecular Endocrine Case Reports". This special issue aims at describing clinical cases in which molecular research was performed, thus opening opportunities for the publication of novel mutations, and for the presentation of clinical particularities in patients with mutations that have already been described. Previous editions of 2008 and 2010 were highly accessed, and had great repercussion. We hope the current edition also fulfills everybody's expectations, or maybe surpasses them.

The advances in molecular genetics methods, in the recent decades, have allowed the identification of genetic components in several endocrinopathies. Many of these discoveries led to important improvements in the diagnosis and treatment of these conditions. One such example is MODY. Alterations in ten different genes have been associated with this disease, and marked differences in the clinical presentation of diabetes caused by mutations in each of these genes were noted. Patients with mutations in *GCK* have mild hyperglycemia, and pharmacological treatment is rarely necessary. In contrast, in individuals harboring mutations in *HNF1A*, hyperglycemia is progressive, and usually requires pharmacological treatment (1,2). This issue presents two studies, by DellaManna and cols. (3) and Caetano and cols. (4), in Brazilian patients with MODY 2 and harboring GCK mutations. Also in relation to the development of hyperglycemia in a monogenic context, a case of maternally-inherited diabetes and deafness (MIDD) with hyporeninemic hypoaldosteronism by Mory and cols. (5) is found in this edition.

In contrast, type 2 diabetes is notably a multifactorial and polygenic disease. The association between polymorphisms in the *TCF7L2* gene and type 2 diabetes have been described in different populations in the recent years, showing a possible role of this gene in the predisposition to the disease. In this edition, such association was investigated in a Brazilian population by Barra and cols. (6).

A number of genetic alterations have been correlated with disorders in growth and sexual development, and in some cases, the genetic profile may predict the phenotype and clinical outcome. From chromosome anomalies to point mutations, studies by Maciel-Guerra and cols. (7), Keselman and cols. (8), Castro and cols. (9), Beneduzzi and cols. (10), Guaragna-Filho and cols. (11), Nishi and cols. (12), and Battistin and cols. (13) concisely address different settings of growth and/or sexual development in which molecular genetics were important tools for a proper clinical approach. Thyroid diseases were also addressed in this issue by Scaglia and cols. (14) in a case of congenital hyperthyroidism, and by Secchi and cols. (15) in a patient with transient congenital hypothyroidism. The study by Geraldo and cols. (16) illustrates the contribution of microRNA detection strategies in the prediction of outcomes, by describing a patient with an aggressive papillary thyroid carcinoma.

In pituitary diseases, better knowledge of the mechanisms involved in tumorigenesis may provide more effective medical therapy. The association of mutations in the *AIP* gene in patients with familial pituitary adenomas has pointed out a possible role of AIP protein alterations in the development of sporadic pituitary tumors. In

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this issue, Kasuki and cols. (17) investigated the AIP expression in GH-secreting tumors in correlation with response to medical treatment. Boguszewski and cols. (18) describe an interesting case in which the two types of multiple endocrine neoplasm coexist, and discuss the genetic findings associated with this condition, whereas multiple endocrine neoplasia type 2 is revisited by Blom and cols. (19) in a description of the rare S891A *RET* mutation in a patient with medullary thyroid carcinoma. In addition, genetic studies in oncogenic osteomalacia by Chang and cols. (20), and in Frasier (Guaragna and cols. [21]), Cowden's (Lima and cols. [22]) and hyperinsulinism/hyperammonemia (Corrêa-Giannella and cols. [23]) syndromes are presented in this issue.

In the past, knowledge about molecular genetics was important in the management of only a restricted group of patients. Currently, this strategy has become critical for proper monitoring of a large number of patients in different settings (24). Accordingly, the practitioner should be familiar with the indications and potential limitations of genetic testing.

We are honored to participate as invited editors in this Special Edition of ABEM, having the opportunity of getting in contact with high-quality articles. We would like to thank the authors who contributed to this edition submitting their manuscripts; the reviewers for their constructive comments and suggestions; and the co-editor of ABEM, Alexander A. L. Jorge, for his invaluable assistance. We hope this edition represents an opportunity to update and acquire new information in such an exciting area of knowledge.

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