Carney complex – case report

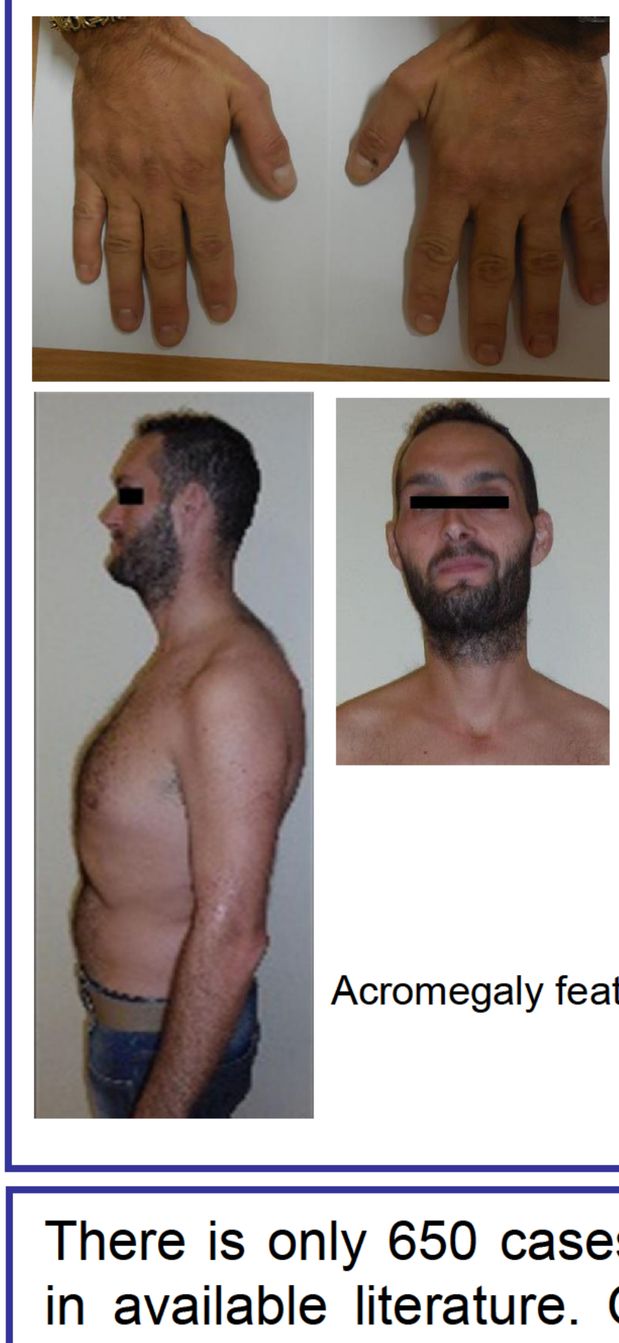
P. Vaňuga[1], M. Kulich[1], H. Urbánková[1], E. Vetešková[1], L. Alexandrová[2]
National institute of endocrinology and diabetology, Lubochna, Slovakia

Introduction

Carney complex (CNC) is rare autosomal dominant disorder, which was firstly described as a combination of manifestation of myxomas, spotty skin pigmentation and endocrine overactivity in 1985. This condition affects many organs and varies in clinical manifestation. The presence of at least two clinical signs is considered as pathognomonic with regard to a sporadic form of CNC, and an evidence of at least one sign with simultaneous manifestation of CNC in any of first degree relatives is considered as pathognomonic with regard to a familiar form. Intracardiac and extracardiac myxomas are the most common diagnostic signs.

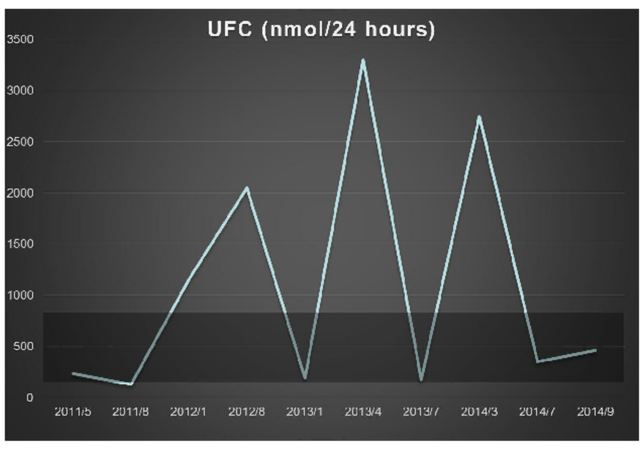
Case report

In our report, we present a case of 36-year male patient with history of surgery for testicular Sertoli – cell tumor and surgery of thyroid nodule, with diagnosis of acromegaly. The patient was examined for persistence of hypersomatotropism after partial resection of growth hormone secreting pituitary adenoma, followed by radiation treatment with Leksell gamma knife and treated by somatostatin analogues.

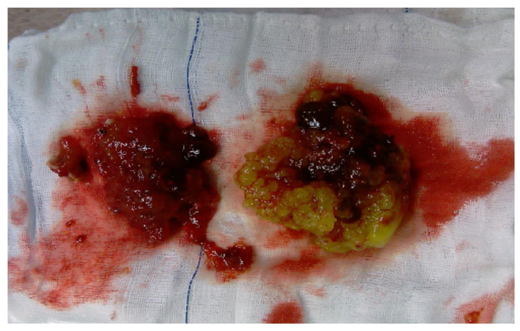


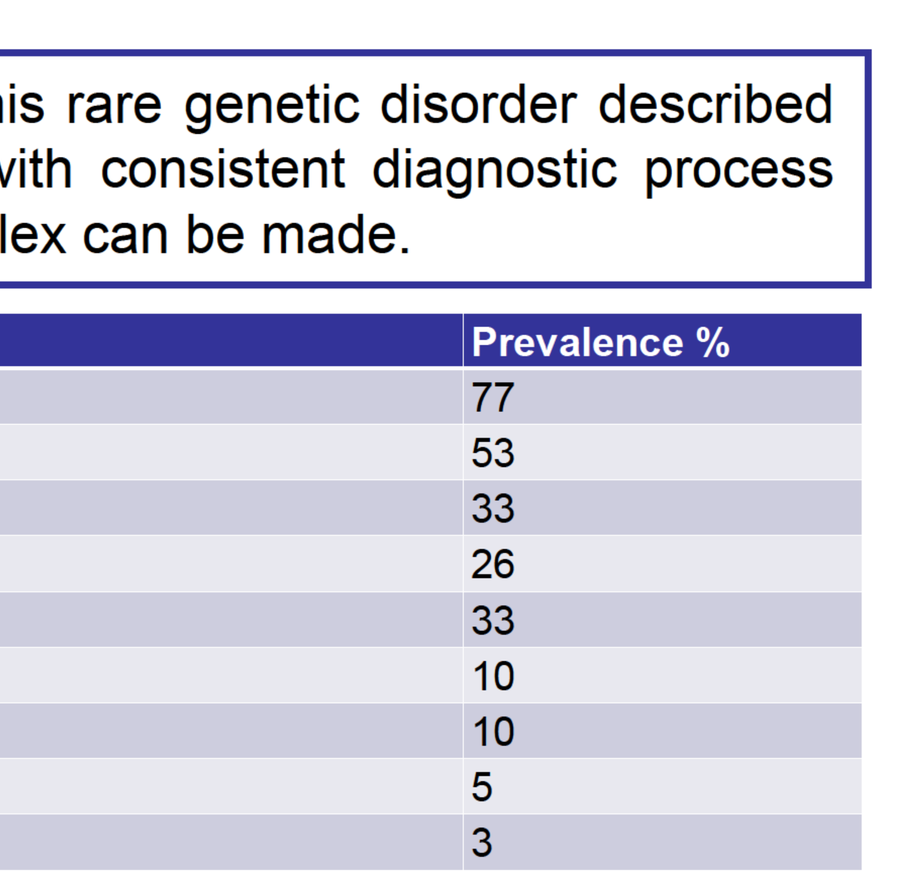
Acromegaly features

Recently, as a part of differential diagnosis procedure aiming to discover other increased endocrine activity, we consider potential occurrence of intermittent hypercortisolism based on primary pigmented nodular adrenocortical disease.



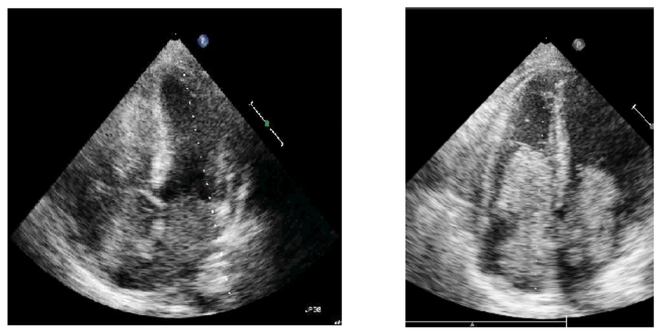
Primary pigmented nodular adrenocortical disease





As a part of comprehensive examination of associated complications of acromegaly we added echocardiography with findings of intra-cardiac tumors. Drifting thrombus, or bacterial flora, or myxomas were considered as options in differential diagnosis. Patient underwent urgent surgical excision of myxomas in left and right atrium at Cardiac Surgery Clinic. The diagnosis of Carney complex was confirmed by genetic testing (deletion of the gene PRKAR1A in nucleotides 491 an 492).

Heart myxomas – echocardiographic findings



There is only 650 cases of this rare genetic disorder described in available literature. Only with consistent diagnostic process the diagnosis of Carney complex can be made.

Clinical features of Carney complex	Prevalence %
Spotty skin pigmentation	77
Heart myxomas	53
Skin myxomas	33
PPNAD	26
Sertolli cell tumor	33
Acromegaly	10
Psamomatous schwanoma	10
Thyroid nodules/ carcinoma	5
Ductal breast adenoma	3

References:
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2. Stratakis CA, kirschner LS. Genetic analysis of Carney complex: current understanding and future guestion. Curr Opin Endocrinol Diab 2002, 9:244-249

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Peter Vanuga
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