

Mutation in *CDKN1B* 3'-UTR region in a patient with acromegaly and primary hyperparathyroidism

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Introduction. Multiple endocrine neoplasia type 4 (MEN4) is a rare disorder, caused by inactivating mutations in *CDKN1B* gene that encodes p27^{kip1} cyclin-dependent kinase inhibitor. To date several different germline *CDKN1B* mutations have been described in patients with clinical features of multiple endocrine neoplasia type 1 (MEN1) negative for *MEN1* mutations (MEN1 phenocopies).

Case report. A female 54 y.o. with clinical features of MEN1, apparently sporadic, negative for *MEN1* germline mutation.

Acromegaly

- Manifestation at 46 y.o.
- Macroadenoma (endo-para(S)-infraselar)
- Residual tissue in left cavernous sinus
After transcranial and transnasal operations

Primary hyperparathyroidism

- Detection at 54 y.o.
- Superior right parathyroid adenoma
- Mild PHPT (serum total Ca - 2.79 mmol/l, iCa - 1,3 mmol/l, PTH - 79 pg/ml)

Other lesions

- Extirpation of uterus and ovaries for adenomatous endometrial polyp, multinodular fibromyoma and endometrioid cyst
- Right mammary gland resection for benign lesion

Genetic testing. High-throughput sequencing on the Ion Torrent Personal Genome Machine (Life Technologies, USA) using a custom-designed AmpliSeq™ panel revealed a heterozygous mutation in 3'-UTR g.3897G>T (c*8G>T) in *CDKN1B* gene. *In silico* testing predicted splice site alterations. Further *in vitro* studies are needed to confirm whether this mutation alters protein function.

Conclusions. To our knowledge, we describe the first mutation in 3'-UTR of *CDKN1B* in a patient with MEN1 phenocopy.

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