Gynecomastia in a patient with von Recklinghausen's disease (Neurofibromatosis type 1).

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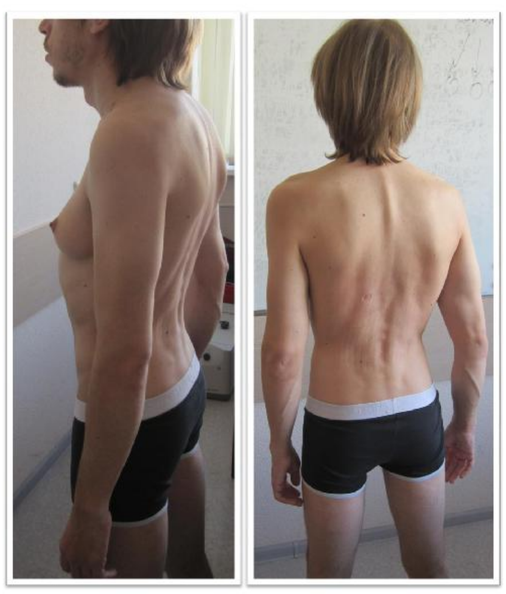
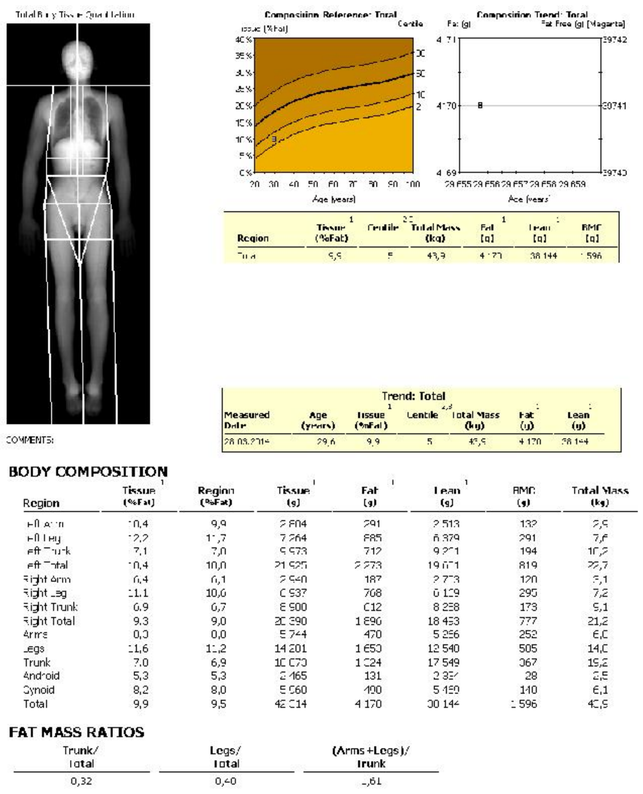
Introduction: Neurofibromatosis type 1 (NF1), also known as von Recklinghausen's disease, is the most common type of NF and one of the most frequent human genetic diseases (prevalence 1:3000). Gynecomastia due to pseudoangiomatous stromal hyperplasia (PASH) in patients with NF1 is quite a rare complication (13 %) but still it should be differentiated from gynecomastia caused by other conditions.

Clinical case: A 30-year old man, previously diagnosed with NF1 at the age of 19, was first consulted in the Endocrinology department of MSMU in March 2014. He complained bilateral enlargement of breasts, painful feeling in them, weight loss, reduction of libido, muscle weakness, back pain and pain in the legs, diarrhea. Physical examination: height 165 cm, weight 42.5 kg, BMI 15.7 kg/m^2, strong reduction of subcutaneous fat, breasts enlarged (Pic 1), painful while palpated; loss of facial hair, multiple neurofibromas in supraclavicular, axillary and parotid areas. Laboratory analysis: serum creatinine 50.8 Mmol/l (62.0-115.0), prolactin 136 mIU/ml (47-135), macroprolactin 61 mIU/ml, estradiol – 97 pmol/l (0-146), testosterone – 9.7 nmol/l (8.4-28.7), leptin – 0.9 ng/ml (2-5.6). DXA: generalized osteoporosis (Z-criteria -3.2 -3.8), Total Body quantitation: total fat tissue – 9.9% (Pic. 2). Inherited lipodystrophy was excluded by genetic testing (16 associated genes were studied). Immunological tests of specific antibodies in celiac disease: negative. Brain MRI: no signs of a pituitary adenoma. Breasts US: gland tissue hyperplasia, ducts are not dilated, no cysts or tumours were detected. Mediastinum lymph nodes biopsy: neurofibromas. Thorax CT (Pic 3): multiple neurofibromas. Spine MRI: multiple neurofibromas. Neurologic status: Multiple compressive-ischemic mononeuropathies of upper and lower limbs, distal assymetric tetraparesis, neuropathic pain syndrome, crampy. ENMG: sensor and motor myelinopathy and axonopathy of upper and lower limbs (Pic. 4).
After 10 months of gluten free diet there are no signs of diarrhea, + 10 kilos weight. Pain syndrome is treated successfully with Pregabalin 375 mg/day and Amitriptyline 0.05 mg/day. A bilateral mastectomy was performed. Osteoporosis is treated with Zoledronic acid and combined preparations of Calcium and Vit D3. Erectile dysfunction was diagnosed and is treated with a 6 weeks course of PDE-5 inhibitor (Tadalafil) with strong positive clinical effect.



Picture 1



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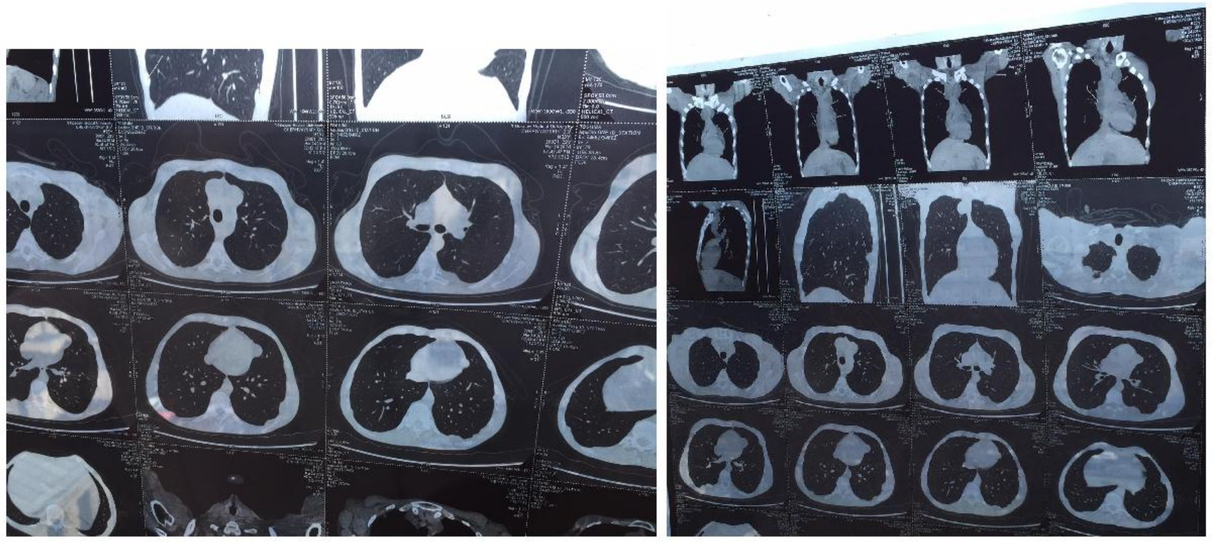
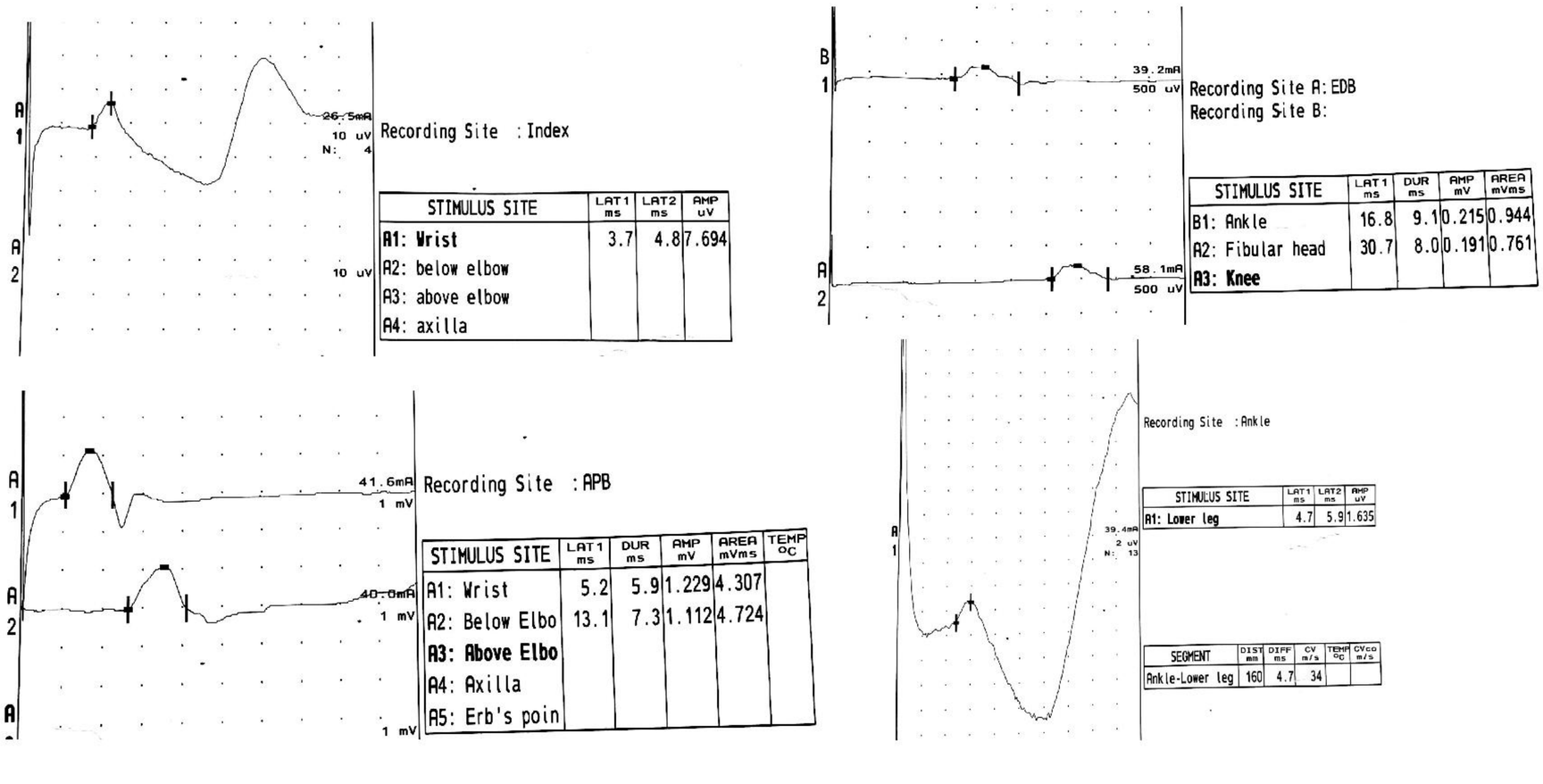


Picture 3



Picture 4

Conclusion

We report our experience of gynecomastia and hyperprolactinemia in a patient with NF1. Though these are rare complications of the disease, if found, such patients should undergo complex examination to exclude other causes of this state, which can worsen the course of their primary disease.

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1282--EP
Clinical Cases - thyroid/others
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DOI: 10.3252/pso.eu.17ece.2015
Poster presented at: ECE2015 16–20 May 2015 DUBLIN, IRELAND 17th EUROPEAN CONGRESS OF ENDOCRINOLOGY