Multiple endocrine disorders in Werner syndrome.

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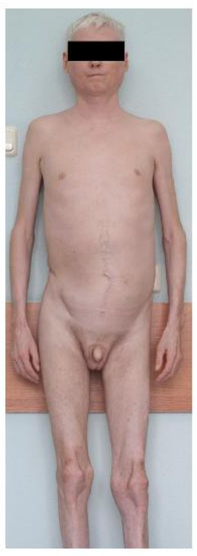
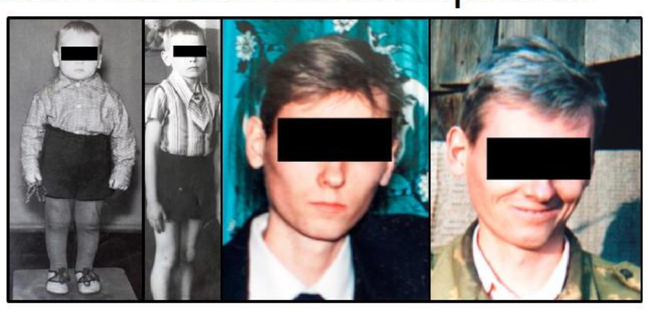
Background: Werner syndrome is a rare autosomal recessive disease caused by a mutation of the DNA helicase gene (*WRN*), characterized by the premature onset of multiple age-related disorders.

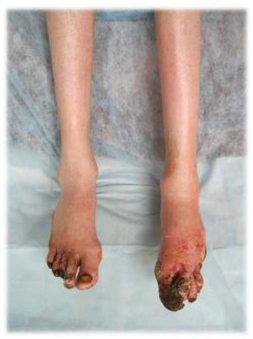
Objective: To describe unusual multiple endocrine and metabolic disorders in 3 unrelated clinical cases of Werner syndrome.

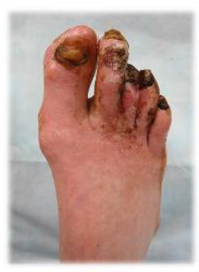
Methods: Three patients with obvious clinical features of premature ageing were referred to an endocrinologist due to endocrine disorders. Mutations of the *WRN* gene were found using a custom Ion Ampliseq panel and PGM semiconductor sequencer (Ion Torrent).

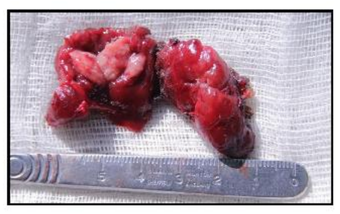
Results: All patients had short stature, grey hair since youth, a beaked nose, BMI deficiency and lipoatrophy, two of them had had their lenses replaced.





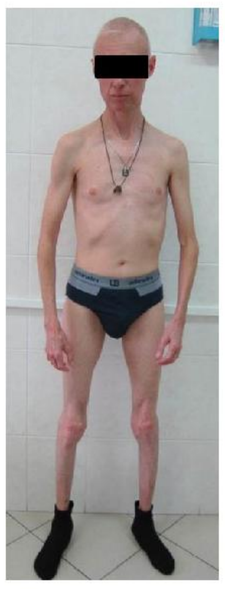




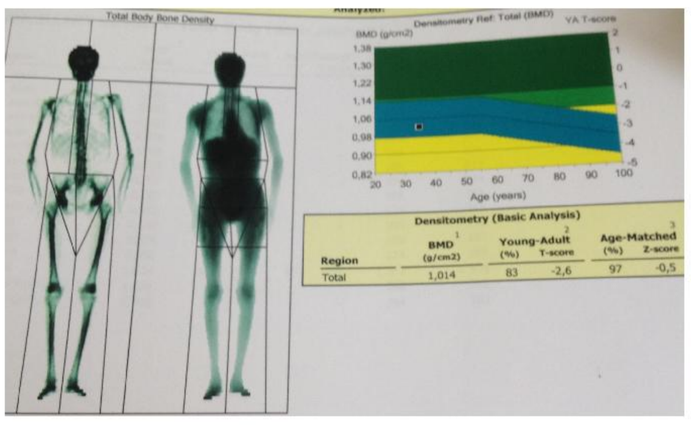


Patient 1

Patient 1, 41 y.o., was referred to an endocrinologist due to uncontrolled diabetes mellitus type 2 (DM2) with marked insulin resistance (200U insulin daily) and late-onset complications (microalbuminuria, polyneuropathy, trophic foot ulcers), marked hypertryglyceridemia and arteriosclerosis, recurrent acute pancreatitis and follicular adenoma of the thyroid gland. In Patient 1 homozygous mutation c.3957dupT p.I1320Y*fs*X12 was found.

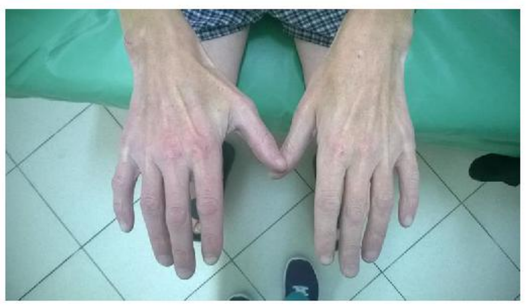


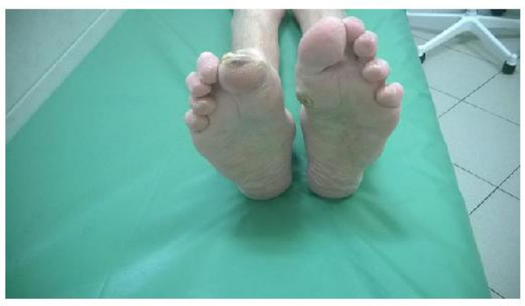
Patient 2



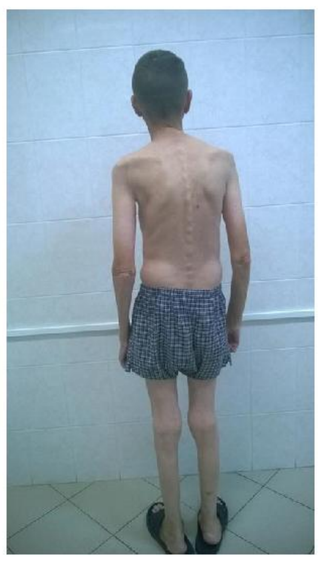
Patient 2, 35 y.o., was referred to an endocrinologist due to hyperparathyroidism, which was secondary to Werner syndrome as no parathyroid tumour was found and vitamin D levels were normal. He had milder DM2 and dyslipidemia, foot ulcers, primary hypothyroidism and osteoporosis. Patient 2 – heterozygous mutation c.1165del. p.R389E*fs*X4.

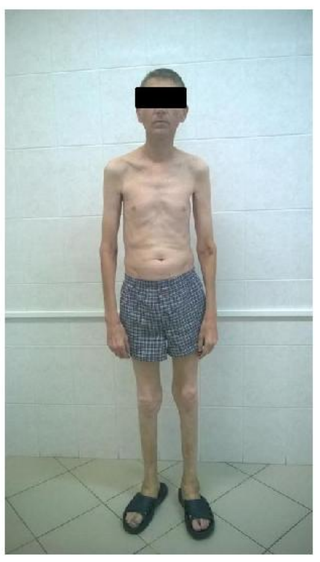
Patient 3, 43 y.o., was referred to an endocrinologist due to severe osteoporosis and hypogonadism, and he also had mild DM2. In Patient 3 heterozygous transition p.L687V (registered as a rare polymorphism rs185468906) was found.





Patient 3





Conclusion

Patients with Werner syndrome develop multiple endocrine and metabolic disorders, which may vary, possibly due to different genetic backgrounds, but this should be recognised and diagnosed by endocrinologists as soon as possible in order to provide patients with required symptomatic therapy and proper follow-up.

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Clinical case reports - Thyroid/Others

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