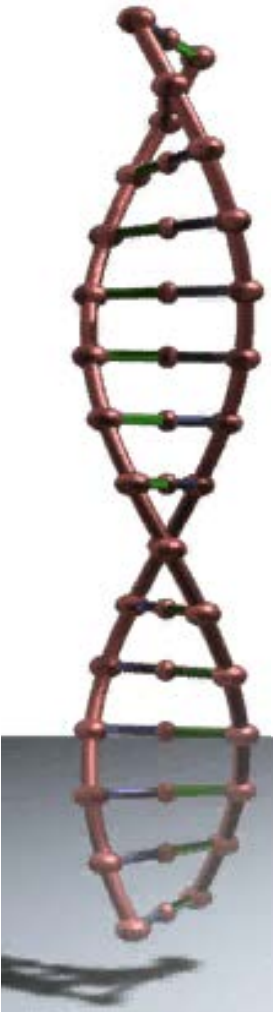

Clinical Case Presentation

Maria Kasesalu

Mirjam Võsaste

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- 47 year old male

- Complaints
  - Progressive proximal muscle weakness for 6-7 years: unable to stand up without the help of hands, incapable of walking uphill
  - For 5-6 years hasn´t been able to run

History

- Fisherman
- Non-smoker
- 2 daughters
- Hypercholesterolaemia
- Type 2 diabetes
- Gynecomasty
- Erectile dysfunction
- Normally built
- Family history: mother and father have type 2 diabetes
- Medication:
  - Olmesartan 10 mg x 1,
  - Metformin 850 mg x 2
  - Gliclazidum 120 mg x 1
  - Nebivololum 2,5 mg x 1
  - Warfarinum
  - Simvastatinum 80 (!) mg x 1

Medical history

- Internal medicine department, 2003
  - Shortness of breath
  - Bihiliary lymphadenopathy – sarcoidosis?
  - Bilateral fluidothorax
  - Cardiomegaly
  - Incidental finding: hoarse systolic murmur
  - Chronic heart failure, NYHA III
  - Echocardiography: severe congenital aortic valve stenosis

Medical history

- Internal medicine department, 2004
  - ACE normal, lymphadenopathy slightly decreased
  - Coronarography: coronary arteries normal
  - Aortic valve prosthesis placement

Laboratory

Laboratory finding	2003	2004	2011	Reference range
ASAT		50	26	<38 u/l
ALAT	52	92	36	<41 u/l
CREA	57	67	54	62-106 µmol/l
CK			336	38-174 u/l
CRP	20	2	1	<5 mg/l
LDH		462	700	240-480 u/l
Chol	5.78	6.33	10.6	<5 mmol/l
HDL-Chol	1.98	2.17	1,47	>1 mmol/l
LDL-Chol	3.48	4.08	5.51	<3.0 mmol/l
Trig		2.17	8.39	<1.7 mmol/l
HbA1C	6.8		normal	%
K	4.6	4.4	5	3.5-5.1 mmol/l
Na		136	139	136-145 mmol/l
Myoglobin			55	28-72 ng/ml

The first hypothesis

- Elevation of CK and transaminase levels are a side effect of a statin use

  NB! Simvastatin on gradually larger doses for 7 years

Neurologists´ consultation

- Proximal muscle weakness
- Walking normal, fast, able to walk on toes, on the soles of feet
- Cranial nerves normal
- Unable to stand up from sitting down, needs the help of his hands
- No atrophies of leg muscles
- Slight atrophies of shoulder muscles
- Tendon reflexes are absent
- Superficial and deep sensation feelings normal
- No neuropathic pains
- Clinical picture: gradually progressive lower motoneuron damage
- Hereditary spinal muscle atrophy?

Electroneuromyography

- Pronounced upper and lower limb neurogenic damage. Significant sensory axonal polyneuropathic damage (lower limbs > upper limbs).

- Proximal muscle weakness (SMA)
- Type 2 diabetes
- Hypercholesterolaemia
- Cardial pathology
- Gynecomasty
- Erectile dysfunction

- Possible diagnoses?
- Further studies?

Genetic testing

- Abnormal CAG insert in the androgen receptor gene → androgen receptor defect
- Expanded trinucleotide repeat (CAG) in the first exon of the androgen receptor gene on the X chromosome
- Normal 10-36 CAG repeats
- Our patient 44 CAG repeats

- Testosteron insensitivity

Kennedy´s disease

- X-linked lower motor neuron disorder
- Progressive weakness of the limb and bulbar musculature
- Often first symptoms: painful cramping, uncontrollable muscle spasms
- Endocrinologic abnormalities: diabetes, testicular atrophy, gynecomastia, oligospermia, erectile dysfunction
- Typically affects men, women can be symptomatic
- Prevalence 1: 40,000 men
- The age of onset - 40-60 years
- Life expectancy normal
- ALS – signs of pyramidal tract disease (spasticity)
- SMA IV – sensory nerve conduction normal
- KD – no spasticity, possible subtle sensory neuropathy

Treatment

- No cure or treatment
- Maintaining maximal function in the presence of this slowly progressive disease
- Stretching exercises for the management of cramps and pain

Once again…

- Type 2 diabetes
- Hypercholesterolaemia
- Cardial pathology
- Muscle weakness
- Gynecomasty

- Take-home message
  - Not uncommon
  - Genetic testing for prognosis
  - Prenatal testing available
  - Careful with statin use

Thank you for listening!

www.kennedysdisease.org