



## The importance of early detection of Adrenomyeloneuropathy (AMN). A case report.

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### OBJECTIVE

**Introduction:** Adrenomyeloneuropathy (AMN) is a form of X-linked adrenoleukodystrophy (X-ALD) a rare inherited disorder (estimated prevalence 1:20,000 to 1:50,000) characterized by the accumulation of very long chain fatty acids (VLCFA) and affects mainly the nervous system white matter and the adrenal cortex and nearly 100% penetrance in males. Widely varying phenotypes and prognoses often co-occur in a single kindred. **Methods:** We report a case diagnosed at age 14 with idiopathic Morbus Addison. He was treated with corticosteroid replacement therapy and was stabilized. At age 27 he was hospitalized in neurology department with right coxo-femoral articulation pain, walking difficulty, progressive weakness and stiffness of the legs and loss of the ability to coordinate muscle movement. In laboratory evaluation routine hemogram resulted normal. Brain and cervical spine MRI resulted normal. Nerve conduction tests revealed a bilaterally significant increase in the central conduction time, four limbs electromyography examination resulted normal. The neurologist diagnosis was a spastic paraparesis (sporadic form) in a subject with morbus Addison. The patient married at age 25 and had 2 children a girl and a boy. The case described above went undiagnosed for almost 10 years until he was sent to our Endocrinology Department at age 37 with worsening of the neurological symptoms. The diagnosis of AMN was established by elevated plasma concentration of VLCFA C26:0, C24:0 and elevated ratio of C24/22 and C26/22. After genetic counseling, blood samples of other family members (mother, sister, brother and children) have been sent for VLCFA analysis. Plasma concentration of VLCFA of his mother, sister and daughter resulted elevated (carriers of X-ALD) but his son and brother resulted normal.

### CONCLUSIONS

Males with idiopathic primary adrenal insufficiency should be evaluated for underlying ALD or AMN. Early diagnosis of AMN helps for detecting the carriers, at-risk female relatives and to decide on prenatal testing for pregnancies at increased risk.

