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Alström Syndrome- a rare disease presenting with dilative cardiomyopathy and blindness. Two case reports.

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Introduction:
Alström Syndrome (AS) is a rare, autosomal recessive single gene disorder with varying clinical features including blindness/ early nystagmus, dilative cardiomyopathy, hearing loss, obesity, Type 2 diabetes mellitus, hepatic dysfunction and renal failure. Diagnosis can be confirmed by analysis of mutations in the ALMS1 gene on chromosome 2p13. Only symptomatic therapy is available. We report on two cases: A 12 year old, blind boy that presented with acute symptoms of cardiac decompensation that led to cardiopulmonary resuscitation and death.Further about a two year old girl with neonatal cardiomyopathy, nystagmus and ventricular septal defect now presenting with severe photophobia during admission for VSD- Repair.

Methods:
Case report, review of literature

Results;
Because of the combination of cardiomyopathy and blindness we suspected Alström syndrome in both cases.
Genetic analysis in both patients revealed mutations in the ALMS1 Gen.

Conclusions:
Neonatal cardiomyopathy in combination with vision impairment and nystagmus are leading symptoms for Alström syndrome. The diagnosis can be confirmed by genetic analysis of the ALMS gene.