Leber Congenital Amaurosis

Description:
- effects 1 in 80,000 births
- about 3,000 live in USA
- gene effected is RPE65
- untreatable
- there are assisting technologies available
- there are resource centers for support and if needed assistant can be assigned to patient

When:
-caused at birth
- is recessive

History:
- discovered byTheodore Leber --> German ophthalmologist
- he found out in 1869 it was inherited defect

Causes:
- missing rods and cones --> work in the retina
- nerve defect

Symptoms:
- vision impairment / blindness
- hearing impaired/ deaf
- mental retardation (rare cases ((20%)))
- decreased physical coordination
-epilepsy (seizing)

Population affecting:
- 5% at effected birth (inborn)
- responsible for 10% of blindness in Sweden and Netherlands

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- no discrimination but certain ethnic groups affected more than

others

Prognosis:
- varies depending of symptoms
- does not effect life expectancy
- Doesn't affect brain, heart, kidney

Children?:
- can have children
    l__> child won't be effected if there are not two of the same type of LCA present whether it be hetero. or homozygous recessive

Interesting Facts:
- even some effected live "normal lives" with boundaries but preserver
- every patient has a specified diagnosis and is 1 of about 6 **different** stages or types.