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Optic atrophy and paraplegia related to Leber's disease

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The Leber's neuropathy is a rare hereditary optic disorder caused by a mutation of mitochondrial DNA in the cytoplasm of the egg. Three primary mutations are often found: 11778, 14484 and 3460. The most common is 11778, it is also the one with the worst visual prognosis. This disease is reflected by an optic atrophy responsible of blindness, transient or permanent depending on the causal genetic mutation. Patient presents a sequential and painless loss of vision in each eye. This neuropathy can be isolated or associated with cardiac damage (Wolff-Parkinson-White's syndrome) or neurological lesions (Leber Plus' disease). We report the case of Leber Plus' disease related to 11778 DNA mutation in a young patient with bilateral optic atrophy and paraplegia.

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