We present the case of an 11-year-old girl with continuously progressive loss of vision. Fundoscopy revealed a picture consistent with Stargardt's disease and fundus flavimaculatus. This article provides a short overview of differential diagnoses worth considering in children and adolescents who present with a similar picture. After our initial workup, genetic chip analysis of the ABCA4 gene was performed, which yielded a heterozygous mutation. Because a second mutation could likely have been on a part of the gene not screened by chip analysis, and in light of the striking picture, a clinical diagnosis of Stargardt's disease was made.
Occurences:
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