Floating-Harbor Syndrome

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Background:

We present a case of a child with Floating-Harbor Syndrome (FHS) with bilateral chorioretinal coloboma (CC). To the best of our knowledge, this is the first case report of this association. Floating- Harbor syndrome is an extremely rare autosomal dominant genetic disorder with approximately 100 cases reported. It is characterized by a series of atypical features that include short stature with delayed bone age, low birth weight, skeletal anomalies, delayed speech development, and dysmorphic facial characteristics that typically portray a triangular face, deep-set eyes, long eyelashes, and prominent nose.

Materials and methods:

Our patient was examined by a pediatric ophthalmologist for the time at age of 7. Visual acuity, optical coherence tomography (OCT) and Optos imaging were collected on every visit. The patient had whole genome sequencing ordered by a pediatric geneticist to confirm Floating-Harbor

syndrome.

Results:

We present the patient's OCT and Optos images that illustrate the location of the patient's inferior chorioretinal coloboma in both eyes. The whole genome sequencing report collected revealed a heterozygous de novo pathogenic variant in the SRCAP gene, consistent with a Floating-Harbor syndrome diagnosis in the literature.

Discussion:

Both genetic and systemic findings are consistent with the diagnosis of Floating-Harbor syndrome in our patient. Rubenstein-Taybi and Floating-Harbor syndrome share a similarity in

molecular and physical manifestations, but because of the prevalence in Rubenstein-Taybi diagnoses, it is a syndromic condition that includes coloboma and frequently associated with each other. Therefore, a retinal exam should become part of the standard protocol for those with FHS, as proper diagnosis, examination and treatment can prevent irreversible retinal damage.