Presentation of Bilateral Optic Disc Coloboma–Morning Glory Syndrome in Mother and Son, with Retinitis Pigmentosa in the Father

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Dear Editor,

Morning glory syndrome (MGS) and optic disc coloboma (ODC) are rare congenital optic disc (OD) anomalies that can be confusing and difficult to differentiate due to their similar presentations. MGS, first described by Kindler in 1970 and named for its resemblance to the morning glory flower, is characterized by a funnel-shaped, enlarged, and excavated OD.[1] Meanwhile, ODC appears as a sharply defined, white excavation within an enlarged OD and can occur in isolation or alongside other ocular anomalies, such as iris or chorioretinal coloboma (CC), particularly in patients with *PAX6* mutations.[2]

Retinitis pigmentosa (RP) is the most common form of inherited retinal degeneration, characterized by progressive photoreceptor disorders leading to photoreceptor cell death.[3] The disease can follow several modes of inheritance, including autosomal dominant, autosomal recessive, and X-linked patterns.[3]

Herein, we highlight a rare familial presentation involving bilateral OD anomalies in a mother and son, as well as CC in the son and RP in the father, and describe the associated genetic findings.

A family consisting of a 44-year-old woman, an 11-year-old boy, and a 40-year-old man visited our clinic. The woman came for a routine examination, while the boy presented with strabismus and the man reported low vision since childhood.

Upon evaluation, the woman's best corrected visual acuity (BCVA) was 20/50 in the right eye and 20/20 in the left. Fundus examination revealed bilaterally excavated, enlarged OD with peripapillary atrophy (Figure 1).

The 11-year-old son's BCVA was 20/25 bilaterally. Cycloplegic refraction revealed a bilateral hyperopic error of +2 diopters (D) in the 90-degree axis. Extraocular motility assessment indicated small-angle esotropia at distance and near fixation (10 prism diopters [PD] and 4 PD, respectively). Slit-lamp fundus examination revealed bilaterally excavated and enlarged ODs surrounded by an annulus of chorioretinal pigmentary deposition. Additionally, an area of chorioretinal atrophy was located between the OD and macula, and CC was observed inferiorly to the OD (Figure 2). His 8-year-old sibling had no detectable ocular or systemic abnormalities.

The visual acuity of the 40-year-old father was counting fingers at 2 meters bilaterally. Slit-lamp examination showed mild bilateral posterior capsular cataract. Fundus examination demonstrated attenuation of the retinal arteries, OD pallor, retinal atrophy, a bull's eye maculopathy pattern, and bone-spicule pigmentation changes, consistent with RP (Figure 3).

Keywords: Morning glory syndrome, optic disc coloboma, retinitis pigmentosa, choroidal coloboma, YAP1, BBS1

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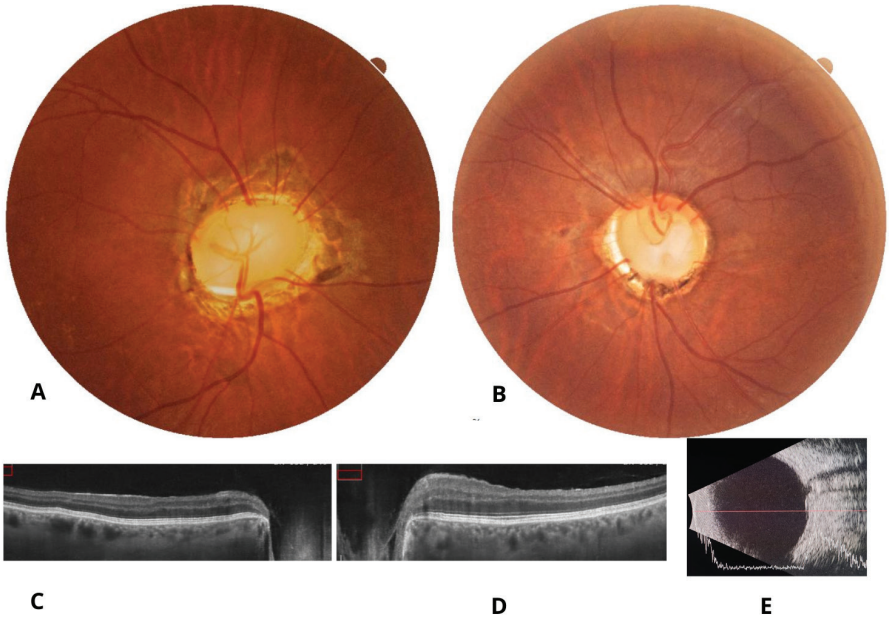

Figure 1. Fundus photographs of the mother's right (A) and left (B) eyes demonstrated bilaterally enlarged, excavated optic discs with surrounding peripapillary atrophy. Macular optic coherence tomography of the right (C) and left (D) eyes revealed significant excavation of the optic disc with preservation of retinal layers, consistent with bilateral optic disc anomaly. The B-scan image of the right eye revealed an excavation in the optic disc (E)

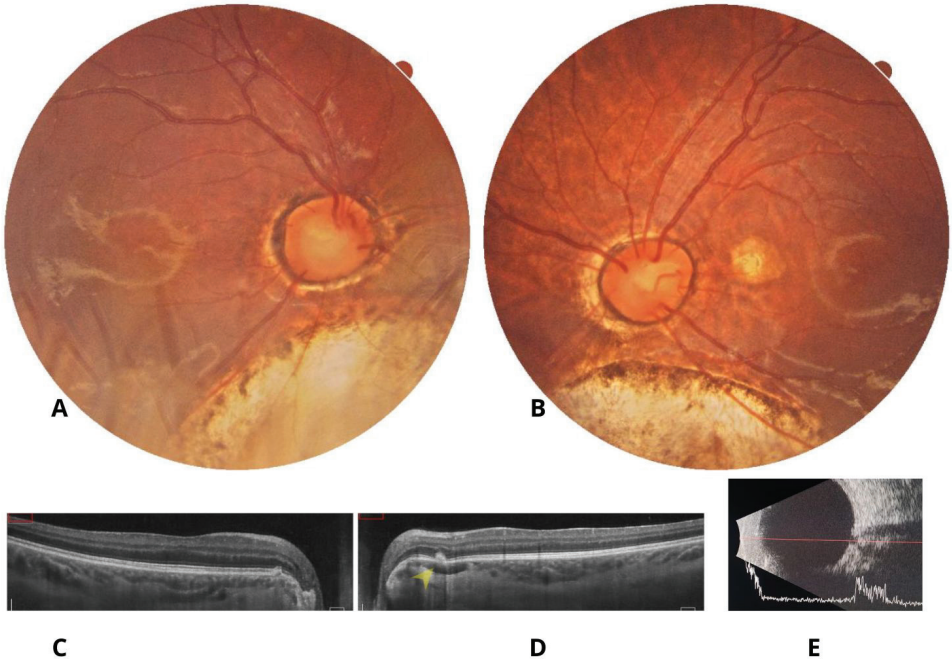

Figure 2. Fundus photographs of the 11-year-old son's right (A) and left (B) eyes showed bilaterally excavated, enlarged optic discs surrounded by an annulus of pigmentary deposition with chorioretinal coloboma inferiorly. Macular optic coherence tomography of the right (C) and left (D) eyes revealed areas of retinal pigment epithelial detachment (yellow arrowhead) with subretinal deposition surrounding chorioretinal atrophy, located between the optic disc and macula. The B-scan image of the right eye illustrated an excavation in the optic disc (E)

To investigate these clinical findings, whole exome sequencing (WES) and targeted variant analysis were performed for the family (Figure 4). The mother and affected son were heterozygous for a variant of uncertain significance in the *YAP1* gene according to the American College of Medical Genetics and Genomics (ACMG) criteria.[4] The unaffected sibling did not carry this variant. Furthermore, the father was homozygous for a pathogenic variant in the *BBS1* gene according to ACMG criteria. However, he exhibited no related systemic manifestations. Both the affected and unaffected sons were also heterozygous carriers of the *BBS1* variant.

Although the exact type of OD anomaly could not be definitively diagnosed, the mother's OD configuration resembled MGS, while the son's was consistent with ODC. The pathogenesis of these conditions differs. MGS may result from a primary mesenchymal abnormality, partial development of the lamina cribrosa, or incomplete closure of the posterior scleral wall, while ODC is caused by defective closure of the embryonic fissure.[1,2] In ODC, the defect is typically decentered inferiorly, reflecting the position of the embryonic fissure and abnormal exit of retinal vessels.[2] In contrast, MGS presents with central excavation and radial exit of the vessels.[2]

YAP1 (OMIM 606608) encodes Yes-associated protein 1, a key effector of the Hippo signaling pathway involved in the development, growth, repair, and homeostasis of multiple organs, including the eye.[5,6] Previous studies have linked *YAP1* variants to various ocular anomalies. DeYoung et al.[5] reported an association with uveal coloboma. Holt et al.[6] described a novel frameshift variant in the *YAP1* gene in a boy with bilateral CC. Meanwhile, Williamson et al.[7] identified novel missense and nonsense *YAP1* variants in families with both syndromic and non-syndromic coloboma. In the present case, both the mother and her affected son carried the identified *YAP1* variant, whereas the 8-year-old son, who exhibited no ocular or systemic abnormalities, did not carry this variant. Unfortunately, segregation analysis could not be extended to the maternal grandparents, as they were deceased. Therefore, the clinical relevance of the identified variant remains uncertain, although it is considered likely to be associated with the observed phenotype based on the available evidence.

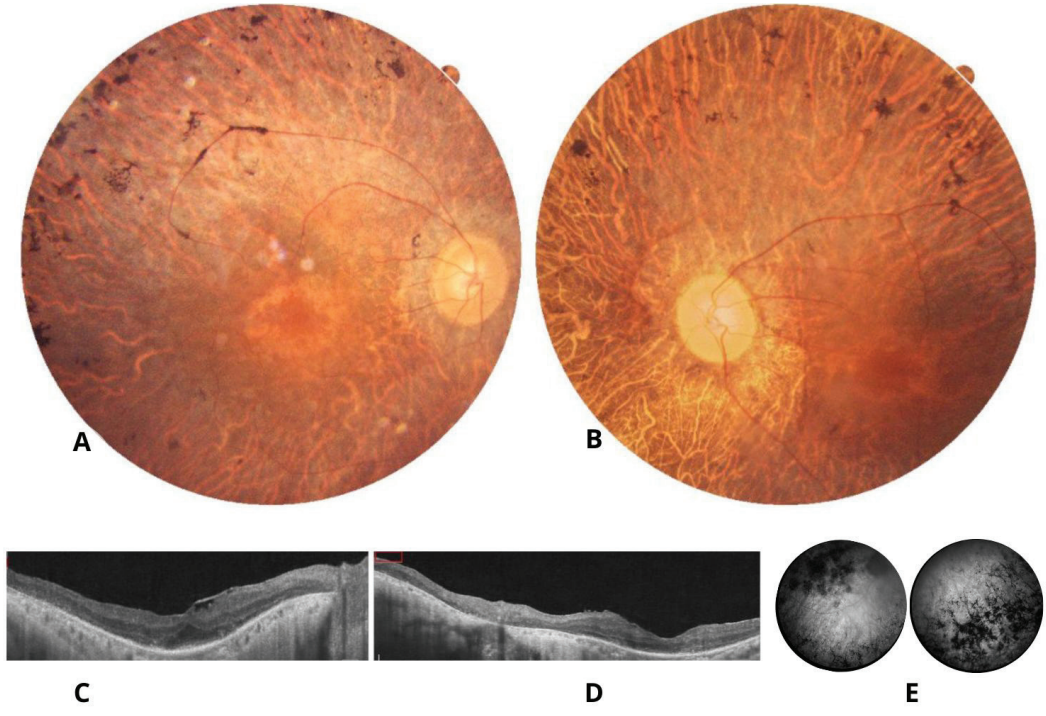

Figure 3. Fundus photographs of the 40-year-old father's right (A) and left (B) eyes revealed features consistent with advanced retinitis pigmentosa, including attenuation of retinal arteries, optic disc pallor, a bull's-eye maculopathy pattern, and extensive retinal atrophy. Macular optic coherence tomography of the right (C) and left (D) eyes demonstrated thinning of the outer retinal layers, loss of the ellipsoid zone, and atrophic changes in the retinal pigment epithelium (RPE). Fundus fluorescein angiography revealed multiple hyperfluorescent window defects observed in the mid-peripheral retina, corresponding to areas of RPE atrophy (E). Pigment clumping in a bone-spicule pattern appears as hypofluorescent areas due to blockage of choroidal fluorescence

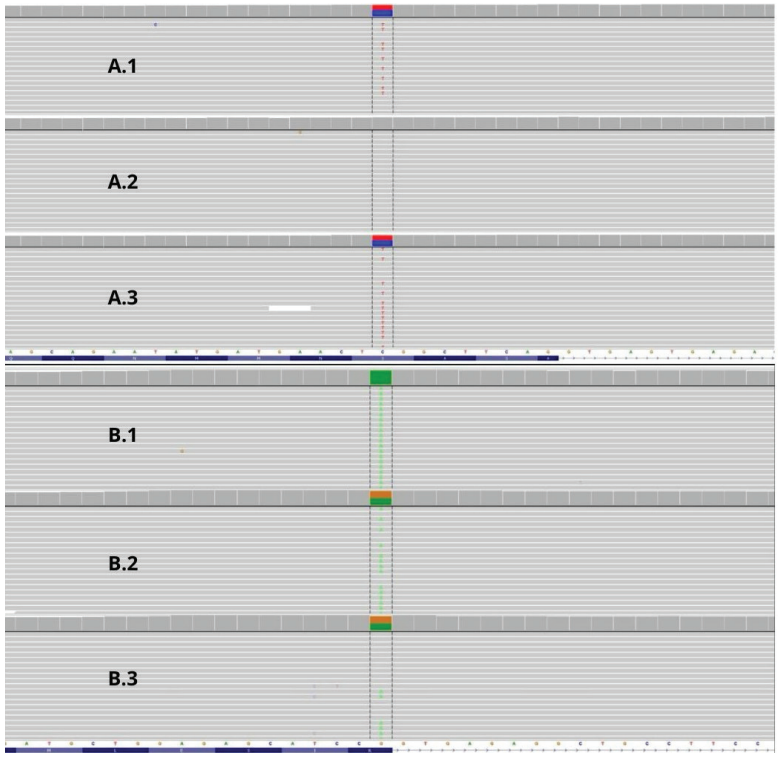

Figure 4. Integrative Genomics Viewer (IGV) images of next-generation sequencing (NGS) data illustrating the genomic region of the *YAP1*(NM_001130145.3):c.680C>T (p.Ser227Leu) variant in the mother (A.1), clinically unaffected son (A.2), and affected son (A.3). The mother and affected son were heterozygous for the variant, while the unaffected son carried the wild-type allele. IGV images of NGS data demonstrate the *BBS1*(NM_024649.5):c.479G>A (p.Arg160Gln) variant in the father (B.1) and his two children (B.2: unaffected son; B.3: son with optic disc anomaly). The father was homozygous for the variant, while both children were heterozygous

Pathogenic variants in the *BBS1* gene associated with RP can present as mild or even nonsyndromic forms of Bardet-Biedl syndrome (BBS).[3] According to WES analysis, the father was homozygous for this *BBS1* variant, whereas the affected son was heterozygous. This difference may partly explain the phenotypic variability observed between the father and son.

Clinically, the CC and OD anomaly observed in the son resembled ODC. This presentation may represent a type 3 fundus coloboma with OD involvement, as described by Gopal et al.[8] In addition, some studies have reported an association between RP and CC, which has been described as an autosomal dominant trait.[9,10] Meanwhile, CC has also been associated with BBS, as reported by Chattannavar et al.,[11] supporting our suggestion of a potential link between the father and son.

The presence of these unusual bilateral findings in the mother and son, both carrying a *YAP1* variant, along with the father's *BBS1*-associated RP, makes this case unique. It underscores the diagnostic challenge of distinguishing between MGS and ODC. Our patients lacked any systemic or neurological abnormalities, particularly cranial midline anomalies. The absence of these associated findings, together with the identification of a heterozygous *YAP1* variant, suggests that the observed phenotype is more consistent with ODC than MGS. Finally, the rare bilateral involvement in two family members raises the intriguing possibility that the son's CC is associated with the father's RP.

Ethics

Informed Consent: Written informed consent was obtained from all participants (and from the parents/legal

guardians for the minor participant) for publication of clinical data and images.

Declarations

Authorship Contributions

Surgical and Medical Practices: Y.İ., B.Ç., Concept: Y.İ., Design: Y.İ., Data Collection or Processing: Y.İ., K.A., Analysis or Interpretation: Y.İ., K.A., Literature Search: Y.İ., Writing: Y.İ.

Conflict of Interest: No conflict of interest was declared by the authors.

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