Scientific Research Publishing

Open Journal of Ophthalmology, 2022, 12, 51-56
https://www.scirp.org/journal/ojoph
ISSN Online: 2165-7416
ISSN Print: 2165-7408

Rhegmatogenous Retinal Detachment in Pierre Robin Anomaly—A Suspicion for Stickler Syndrome: Case Report

Maryam A. Hadi Almohsen*, Fatema Bin Rajab

Salmanyia Medical Complex, Manama, Kingdom of Bahrain
Email: *malmohsen80@gmail.com

How to cite this paper: Almohsen, M.A.H. and Rajab, F.B. (2022) Rhegmatogenous Retinal Detachment in Pierre Robin Anomaly—A Suspicion for Stickler Syndrome: Case Report. *Open Journal of Ophthalmology*, **12**, 51-56.
https://doi.org/10.4236/ojoph.2022.121006

Received: December 26, 2021
Accepted: February 7, 2022
Published: February 10, 2022

Abstract

Stickler syndrome (SS) is an autosomal dominant inherited genetic disorder that presents with hearing loss, a cleft palate, epiphyseal dysplasia, and degeneration, similar to arthritis and well known to be associated with rhegmatogenous retinal detachments. A particular group of physical features called Pierre Robin sequence is also common in people with stickler syndrome. Pierre Robin sequence includes a cleft palate, glossoptosis, and micrognathia. We describe a case report of a family diagnosed with stickler syndrome presenting with Pierre Robin sequence and share some universal management steps for rhegmatogenous retinal detachment in stickler syndrome. Genetic testing is important to support the diagnosis and conduct screenings of family members.

Keywords

Retinal Detachment, Scleral Buckling, Pathological Myopia, Retinal Breaks

1. Introduction

Stickler syndrome is an inherited collagenopathy that was first described by Gunnar Stickler in 1956 as "hereditary progressive arthro-ophthalmopathy" [1]. It is associated with the defective formation of collagens II, IX and XI [2] [3] [4], therefore presenting with a wide range of manifestations namely ophthalmic, musculoskeletal, craniofacial and auditory [4] [5]. Often, stickler syndrome is diagnosed first by the ophthalmologist. Ophthalmic manifestations include moderate to high myopia, hypoplastic vitreous, and a high rate of retinal detachment [3] [4] that can be bilateral [6] [7] [8]. Retinal detachment is a cause of blindness at the young age group between 10 - 30 years [4] [8] and requires multiple sur-

gical interventions due to the higher rate of recurrence leading to a poor final visual outcome in a significant percentage of patients [4] [8] [9]. Other ophthalmic manifestations include congenital cataracts and glaucoma-related to congenital abnormalities of the angle [3] [4] [10]. Orofacial manifestations are variable clinically even within the same family presumably having the same genetic variant [11]. These include midfacial hypoplasia, depressed nasal bridge, long philtrum and mid-line clefting with or without Pierre Robin sequence, which is the triad of micrognathia, glossoptosis and airway obstruction [4] [5] [11]. Clinical diagnostic criteria based on the scoring system for stickler syndrome have been proposed, but not yet validated as a tool of established diagnosis [4] [12] [13]. The gold standard of diagnosis is genetic confirmation. Six identified variants for the disease: COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3. All are inherited in an autosomal manner, variants COL2A1, COL11A1, or COL11A2 being dominant and COL9A1, COL9A2, or COL9A3 being recessive [13]. The classic Type I stickler syndrome is related to COL2A1 and inherited in an AD mode.

2. The Case

An 11 year old boy presents with an acute history of painless total loss of vision for two days duration in his left eye. His fundus exam revealed a 270-degree involved retinal detachment with a dramatic folding of the retina (**Figure 1**). Upon further history, he denied any trauma to the eye. There is a family history of myopia present in his mother and younger brother. Significant past history confirmed that he was given a preliminary diagnosis of "Pierre Robin syndrome" where he and his younger brother had multiple surgeries after birth for cleft palates and retrognathia (**Figure 2** and **Figure 3**). He otherwise was raised healthy and was performing well in school. The mother and father deny any sanguinity, they have 4 children, only two of whom were myopes which had congenital palate and mouth anomalies. The mother gave a history of congenital cataract in her childhood and keratoconus in the current time. Both children have micrognathia and glossoptosis.

After an in-depth discussion with the parents about the need for surgery, they decided to proceed. He was operated two days later with scleral buckle, vitrectomy, endolaser and silicon oil. Intraoperatively there appeared to be a subtotal dialysis except for two clock hours at 6 o'clock with empty vitreal cavity. An indented retinal examination of the right eye showed areas of white without pressure and anomalous vitreous attachments hence barricade laser was done in that eye. The patient was doing well postoperatively with an improvement of vision to 6/36. At 8 weeks he was reoperated for removal of peripheral tractional bands which had developed as a proliferative vitreoretinopathy sequel with some laser augmentation (**Figure 4**).

The younger brother was examined under anesthesia a few days later and was found to be having dense cortical cataract, high intraocular pressure of 45 and 50

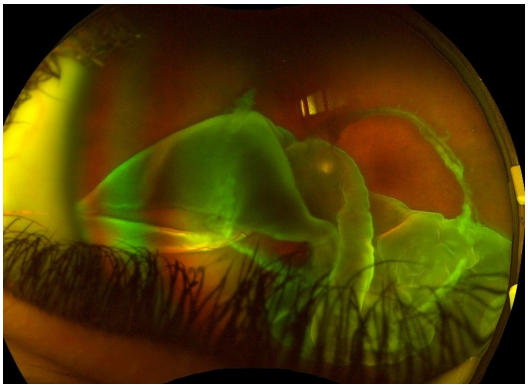

Figure 1. 270 degree retinal break with retinal folding in the patients left eye.

Figure 2. The patient at birth with prominent micrognathia.

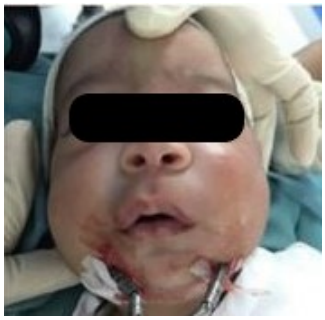

Figure 3. The patient after multiple surgical repairs to the jaw anomalies.

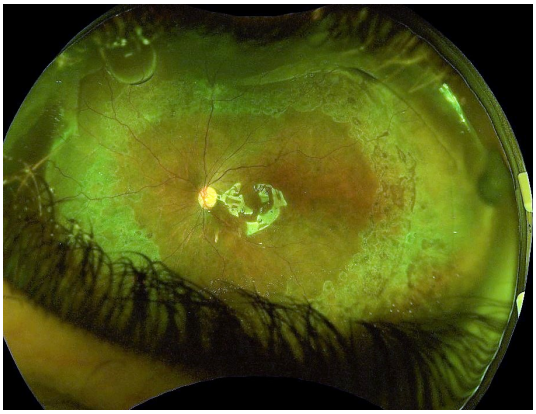

Figure 4. postoperative fundus image of the left eye under silicon oil.

mmhg in the right and left eye respectively. In addition, peripheral 360 laser retinopexy was performed due to the same peripheral retinal changes of vitreous condensation just posterior to the vitreous base and slight discolorations between the base and equator in all quadrants. On follow up his eye pressure showed good response to topical IOP reducing medications.

Based on the retinal findings in both children there was a high suspicion of stickler syndrome so the family was offered genetic testing which revealed positive stickler related mutation of COL2A1 in the patient, the younger sibling and the mother.

3. Discussion

Stickler syndrome is clinically diagnosed with genetic confirmation. Since the syndrome can present with a wide range of manifestations including different systems of the body a high clinical suspicion should be kept when the patient identified to have any of the established clinical signs. The incidence of SS is estimated at 1 in 7500 newborns.

Pierre Robin Sequence (PRS) is a rare congenital anomaly with an incidence of 1/30,000 [14]. The syndrome consists of the triad of micrognathia, glossoptosis and airway obstruction (mostly posterior cleft of the secondary palate). It can occur in isolation or as a part of a more complex syndrome, one of these examples is SS. The diagnosis of PRS in the context of SS is rarely established by the plastic surgeon when the baby is diagnosed PRS after birth as eye manifestations are not yet manifested. The incidence of PRS in SS is rare as well. Moreover, the incidence of visually threatening complications of SS within the PRS sequence is rarely reported [15]. In 2007 Chan *et al.* studied 91 patients with isolated cleft of the secondary palate, only 8 patients (9%) manifested the classic PRS. Among these 8 patients, only 3 (3.2%) diagnosed with SS.

In our case, we are reported two siblings of the same family proven genetically with this rare variant of the disease. Both siblings were diagnosed clinically with PRS after birth, and both were operated for cleft palate. Later when one of the siblings presented to our facility with a giant retinal tear and RRD with retinal folding, the suspicion of SS was raised. Both siblings had high myopia (mention numbers here), visually insignificant cataract and sibling 2 had high intraocular pressure and peripheral retinal changes. The mother was examined as well found to have high myopia and visually insignificant cataract.

The three family members were sent for genetic evaluation and the presence of COL2A1 variant with PRS was confirmed.

4. Conclusion

The case report supports the high prevalence of rhegmatogenous retinal detachment in stickler syndrome cases and the rare possibility of a Pierre Robin sequence to be manifested in such families. Genetic testing is important to confirm the diagnosis and facilitate screening of future generations and family planning.

The need for buckle surgery and laser augmentation should be considered and regular follow-up examinations to rule out recurrence of detachment are usually required.

Acknowledgements

Clinical investigator Dr. Mariam Abu Suhail conducted the genetic testing on the family in the case report.

Consent

As the patient is a minor, the consent has been gathered from the legal guardians; in this case, the parents agree to publish this case and images.

Conflicts of Interest

All authors declare that there is no conflict of interest with the publication of this case report.

References

[1] Stickler, G.B., Belau, P.G., Farrell, F.J., Jones, J.D., Pugh, D.G., Steinberg, A.G. and Ward, L.E. (1965) Hereditary Progressive Arthro-Ophthalmopathy. *Mayo Clinic Proceedings*, **40**, 433-455.

[2] Snead, M.P. and Yates, J.R. (1999) Clinical and Molecular Genetics of Stickler Syndrome. *Journal of Medical Genetics*, **36**, 353-359.

[3] Snead, M.P., McNinch, A.M., Poulson, A.V., Bearcroft, P., Silverman, B., Gomersall, P., Parfect, V. and Richards, A.J. (2011) Stickler Syndrome, Ocular-Only Variants and a Key Diagnostic Role for the Ophthalmologist. *Eye*, **25**, 1389-1400. https://doi.org/10.1038/eye.2011.201

[4] Boothe, M., Morris, R. and Robin, N. (2020) Stickler Syndrome: A Review of Clinical Manifestations and the Genetics Evaluation. *Journal of Personalized Medicine*, **10**, 105. https://doi.org/10.3390/jpm10030105

[5] Stickler, G.B., Hughes, W. and Houchin, P. (2001) Clinical Features of Hereditary Progressive Arthro-Ophthalmopathy (Stickler Syndrome): A Survey. *Genetics in Medicine*, **3**, 192-196. https://doi.org/10.1097/00125817-200105000-00008

[6] Shapiro, M.J., Blair, M.P., Solinski, M.A., Zhang, D.L. and Jabbehdari, S. (2018) The Importance of Early Diagnosis of Stickler Syndrome: Finding Opportunities for Preventing Blindness. *Taiwan Journal of Ophthalmology*, **8**, 189-195. https://doi.org/10.4103/tjo.tjo_97_18

[7] Poulson, A.V., Hooymans, J.M., Richards, A.J., Bearcroft, P., Murthy, R., Baguley, D.M., Scott, J.D. and Snead, M.P. (2004) Clinical Features of Type 2 Stickler Syndrome. *Journal of Medical Genetics*, **41**, e107. https://doi.org/10.1136/jmg.2004.018382

[8] Coussa, R.G., Sears, J. and Traboulsi, E.I. (2019) Stickler Syndrome: Exploring Prophylaxis for Retinal Detachment. *Current Opinion in Ophthalmology*, **30**, 306-313. https://doi.org/10.1097/ICU.0000000000000599

[9] Ang, A., Poulson, A.V., Goodburn, S.F., Richards, A.J., Scott, J.D. and Snead, M.P. (2008) Retinal Detachment and Prophylaxis in Type 1 Stickler Syndrome. *Ophthalmology*, **115**, 164-168. https://doi.org/10.1016/j.ophtha.2007.03.059

[10] Nielsen, C.E. (1981) Stickler's Syndrome. *Acta Ophthalmologica* (*Copenhagen*), **59**, 286-295. https://doi.org/10.1111/j.1755-3768.1981.tb02991.x

[11] Jackson, O.A., Kaye, A.E., Lee, A., Minugh-Purvis, N., Cohen, M.A., Solot, C.B., McDonald-McGinn, D., Jawad, A.F., Zackai, E.H. and Kirschner, R.E. (2020) Orofacial Manifestations of Stickler Syndrome, an Analysis of Speech Outcome and Facial Growth after Cleft Palate Repair. *Annals of Plastic Surgery*, **84**, 665-671. https://doi.org/10.1097/SAP.0000000000002114

[12] Rose, P.S., Levy, H.P., Liberfarb, R.M., Davis, J., Szymko-Bennett, Y., Rubin, B.I., Tsilou, E., Griffith, A.J. and Francomano, C.A. (2005) Stickler Syndrome: Clinical Characteristics and Diagnostic Criteria. *Am. Journal of Medical Genetics A*, **138**, 199-207. https://doi.org/10.1002/ajmg.a.30955

[13] Robin, N.H., Moran, R.T. and Ala-kokko, L. (2021) Stickler Syndrome Summary Genetic Counseling Suggestive Findings. 1-19.

[14] Zheng, W., Li, B., Zou, Y. and Lou, F. (2019) The Prenatal Diagnosis and Classification of Cleft Palate: The Role and Value of Magnetic Resonance Imaging. *European Radiology*, **29**, 5600-5606. https://doi.org/10.1007/s00330-019-06089-9

[15] Huang, F., Kuo, H.K., Hsieh, C.H., Lai, J.P. and Chen, P.K.T. (2007) Visual Complications of Stickler Syndrome in Paediatric Patients with Robin Sequence. *Journal of Cranio-Maxillofacial Surgery*, **35**, 76-80. https://doi.org/10.1016/j.jcms.2007.01.001