

A patient with Stickler syndrome in Pregnancy

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Stickler syndrome is a connective tissue disorder which is clinically suspected but genetically diagnosed. It is characterised by ocular findings (retinal detachment, myopia and cataract), hearing loss (conductive and sensorineural), cleft palate, midfacial underdevelopment, spondyloepiphyseal dysplasia and arthritis. It is autosomal dominant, if the pathological variant is COL2A1, COL11A1 or COL11A2. However, it is autosomal recessive, if the pathogenic variant is COL9A1, COL9A2, or COL9A3.

Case report:

A 33-year-old lady G1 P0 who has stickler syndrome with COL2A1 gene alteration, with a 50% chance of penetrance presented for booking at 12 weeks, her BMI was 41.

She has a prosthetic eye (previous retinal detachment and consequently, the eye was removed) and laser therapy to the right eye to stabilize the retina. She had bilateral hip arthritis and high arched palate.

She has been managed by multidisciplinary team of obstetricians, ophthalmologists, anaesthetists and genetic counsellors. The patient declined referral to foetal medicine.

Management:

In view of the risk of retinal detachment, she had been advised against vaginal delivery by the ophthalmology team as there is a risk of blindness.

She had a CS delivery at 38/40 with steroids for a breach baby. It was uneventful procedure with no postnatal complications.

Following birth, the baby had an ophthalmic review and hearing test, and genetic testing was offered.

Conclusion:

A multidisciplinary team using a patient centred approach is a key for a favourable outcome for patients with uncommon medical conditions during pregnancy and labour.

Autosomal Dominant Inheritance Pattern

