

## Awareness Of Stickler Syndrome Among Dental Students

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### Abstract

#### Introduction

Stickler syndrome is a genetic disorder that can cause serious vision, hearing and joint problems. Also known as hereditary progressive arthro-ophthalmopathy, Stickler syndrome is usually diagnosed during infancy or childhood. Children who have Stickler syndrome often have distinctive facial features — prominent eyes, a small nose with a scooped-out facial appearance and a receding chin. They are often born with an opening in the roof of the mouth (cleft palate). While there is no cure for Stickler syndrome, treatments can help control symptoms and prevent complications. In some cases, surgery may be needed to correct some of the physical abnormalities associated with Stickler syndrome.

#### Materials and methods

An online survey questionnaire was circulated among 100 dental students of saveetha dental college. The data was compiled in Excel and the results were statistically analysed using spss software.

#### Results

From the data analysed, the results showed that 74% of the UG students know about stickler syndrome, and 26% of the PG students know about stickler syndrome, but they are not aware about the cause, complication, and treatment for stickler syndrome.

## Conclusion

From the study it is clear that the students are only aware about the term stickler syndrome but they are not aware about the characteristic feature and other factors related to the syndrome.

## Keywords

Awareness, students, dental, stickler syndrome

## INTRODUCTION

Stickler syndrome, also known as hereditary progressive arthro-ophthalmopathy (ORPHA828), is a connective tissue illness that affects 1 in every 7,000 to 9,000 babies. Stickler syndrome is characterised by a relatively flattened facial look. Underdeveloped bones in the center of the face, such as the cheekbones and the bridge of the nose, give this appearance(1). In patients with Stickler syndrome, a series of physical characteristics known as the Pierre Robin sequence is also common. A cleft palate, a tongue that is positioned further back than normal (glossoptosis), and a tiny lower jaw are all part of the Pierre Robin sequence (micrognathia). This combination of characteristics might cause feeding issues as well as breathing difficulties. Nearsightedness is common in patients with Stickler syndrome (high myopia). The clear gel that fills the eyeball (the vitreous) might have an irregular appearance in some situations, which can be seen during an eye examination. Other eye issues include glaucoma (increased pressure within the eye), cataracts (clouding of the lens of the eyes), and tears of the eye lining (retinal detachment). In some circumstances, these eye defects result in decreased vision or blindness.

Hearing loss in patients with Stickler syndrome varies in severity and might worsen with time. The hearing loss may be sensorineural, meaning it is caused by alterations in the inner ear, or conductive, meaning it is caused by middle ear disorders. Stickler syndrome is characterised by skeletal abnormalities that affect the joints in the majority of patients. Affected children and young adults' joints may be loose and flexible (hypermobile), yet as they become older, joints become less flexible. Arthritis can strike at any age and cause joint pain and stiffness. Problems

with the bones of the spine (vertebrae), such as improper spine curvature (scoliosis or kyphosis) and flattened vertebrae, can also arise (platyspondyly). Spinal discomfort may be caused by certain spinal disorders.

Stickler syndrome is split into numerous categories based on the genetic collagen deficiency that causes it. Patients with autosomal dominant Stickler syndrome have been discovered to have abnormalities in three separate collagen genes(2). Type I Stickler syndrome (STL1) is linked to mutations in the COL2A1 gene, which codes for type II collagen, whereas type II (STL2) and type III (STL3) are linked to mutations in the COL11A1 and COL11A2 genes, which code for type XI collagen, respectively. Mutations in the type IX collagen genes COL9A1 (STL4) and COL9A2 (STL5) have been linked to autosomal recessive Stickler syndrome in some families(3). To some extent, phenotypic differentiation between patients with mutations in different causal genes is conceivable. Because COL11A2 is not expressed in the vitreous, STL3 does not show ocular defects. The vitreous anomaly, which is primarily 'membranous' in STL1 and 'beaded' in STL2, is another example. However, there is still a significant phenotypic difference that cannot be explained entirely by the damaged gene. Clinical manifestation is highly variable even within one family or between unrelated families bearing the same mutation (4–12), (13–18), (19–25)

Stickler et al. documented a family with combined symptoms and increasing myopia, which was linked to retinal detachment and blindness in the first decade of life. Mild sensorineural hearing impairment was included in the symptoms of this disease in a separate report. Hearing loss has since become a common symptom of Stickler syndrome, despite the lack

of precise descriptions and the fact that few investigations have focused solely on the auditory phenotype(26). Hearing loss appears to be present in roughly 60% of STL1 patients and is most likely sensorineural. Hearing loss occurs more frequently and is more severe in those with STL2 and STL3. The pathophysiology of sensorineural hearing loss remains a mystery. A hypermobile tympanic membrane and cleft palate resulting in middle ear effusion and conductive hearing loss are associated findings(27).

Patients with a cleft palate or cleft lip often require complex long term orthodontic treatment, often in combination with a number of other specialists including maxillofacial surgery in order to produce a good facial appearance, with an aesthetic, functional and stable occlusion. So Dentists must be aware and must be able to identify and differentiate if the individual has stickler syndrome or not(28). So, the main aim of the study is to check if the dental students are aware of what stickler syndrome is and about its associated features.

## MATERIALS AND METHOD

To analyse the knowledge and awareness of stickler syndrome among dental students, an online survey was conducted in a standard manner by simple random sampling method involving all age groups, by distributing 15 questionnaires (given below) through an online portal called Google forms, among 200 dental practitioners including interns dental postgraduates and dentist from different locations by acquiring approval from the ethical committee and proper consent from the individuals involving in filling the survey and then the stratified data was collected and then data manipulation was done by clean up in an excel spreadsheet and the result was collected and then analysed through SPSS software. Descriptive statistical analysis and Chi Square test for comparison of parameters were used and P value is calculated.

## RESULT

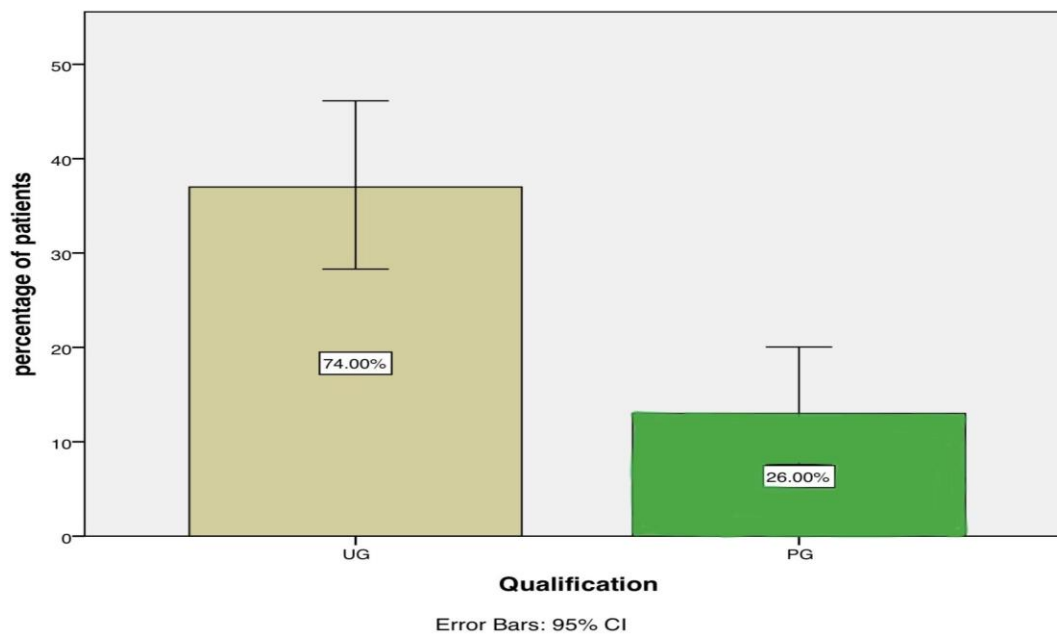


Figure 1 - This figure shows a bar graph between Qualification and the number of participants. The brown colour represents UG students, and the green colour represents PG students. The graph shows that 74% of the UG students have

participated in the survey and 26% of the PG students have participated in the survey. The chi square test was evaluated for this graph and obtained p value of  $p = 0.316$  ( $p > 0.05$ ). Hence the value is statistically insignificant

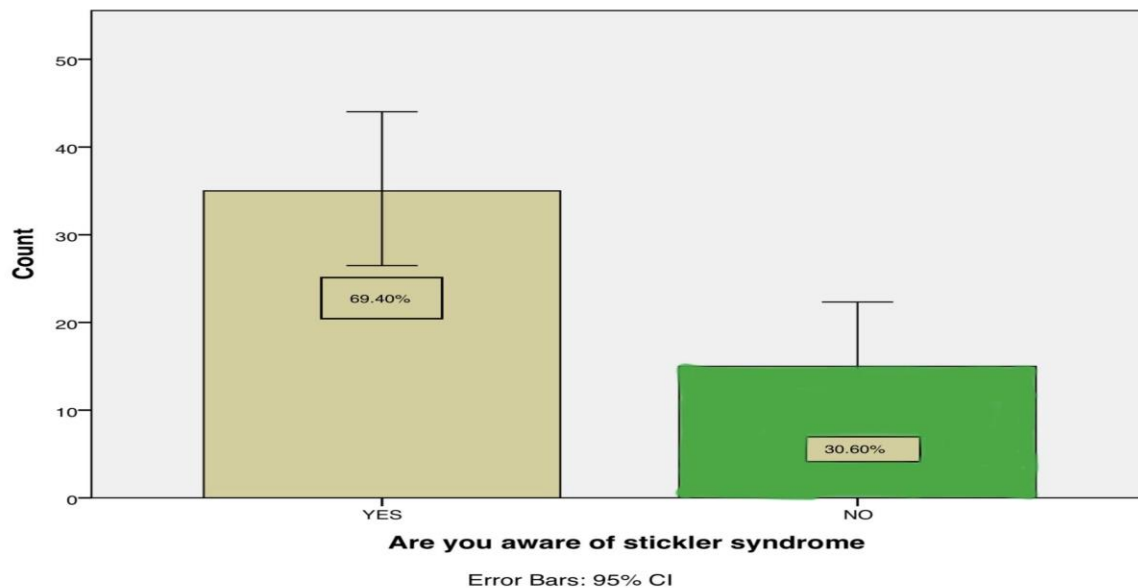


Figure 2 - This figure shows a bar graph between the number of participants against how much they are aware of what stickler syndrome is. The brown colour denotes yes, and the green colour denotes no. The graph shows that 69.40% of the participants know what stickler syndrome is and

30.60% of the participants does not know what stickler syndrome is. The chi square test was evaluated for this graph and obtained p value of  $p = 0.142$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

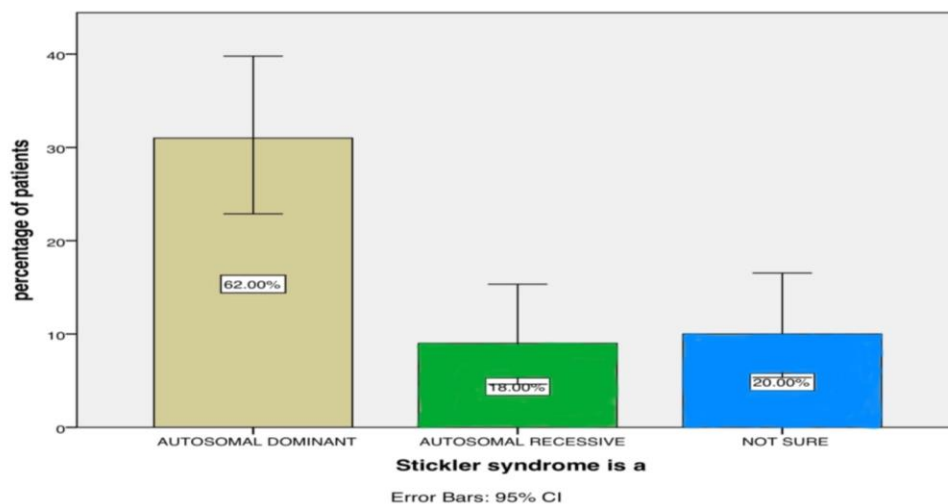


Figure 3 - This figure shows a bar graph if stickler syndrome is autosomal dominant or autosomal recessive or if they are not sure against if they are aware among the number of participants. The brown colour represents autosomal dominant, the green colour represents autosomal recessive

and the blue colour represents that they are not sure. From the graph 62% of the participants said stickler syndrome is a autosomal dominant disorder and 18% of the participants said that stickler syndrome is autosomal recessive and 20% of the participants are not sure. The chi

square test was evaluated for this graph and obtained p value of  $p = 0.032$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

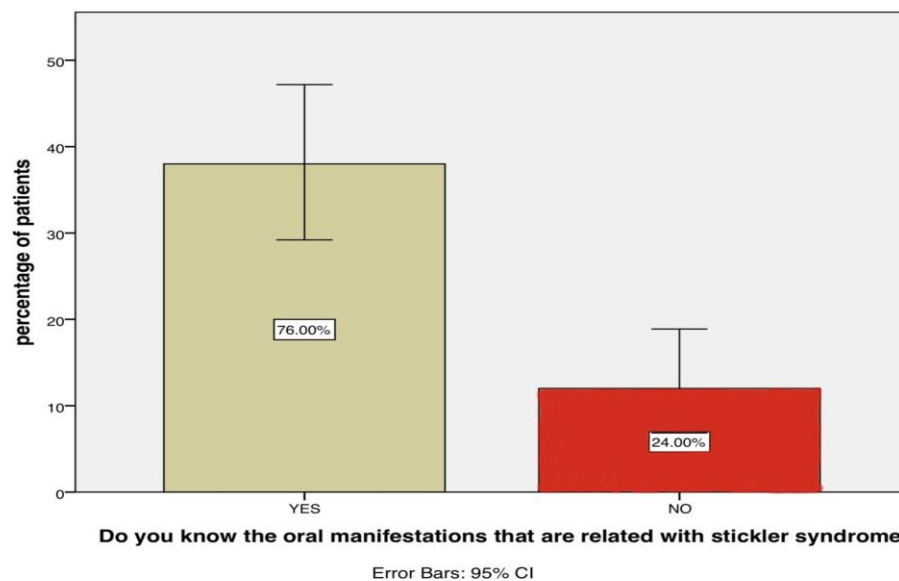


Figure 4 - This figure shows a bar graph about the oral manifestation of stickler syndrome against the awareness of the participants. The brown colour represents yes, the red colour represents no. From the graph 72% of the participants said that they are aware about the oral

manifestation and 24% of the participants are not aware about the oral manifestation of stickler syndrome. The chi square test was evaluated for this graph and obtained p value of  $p = 0.245$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

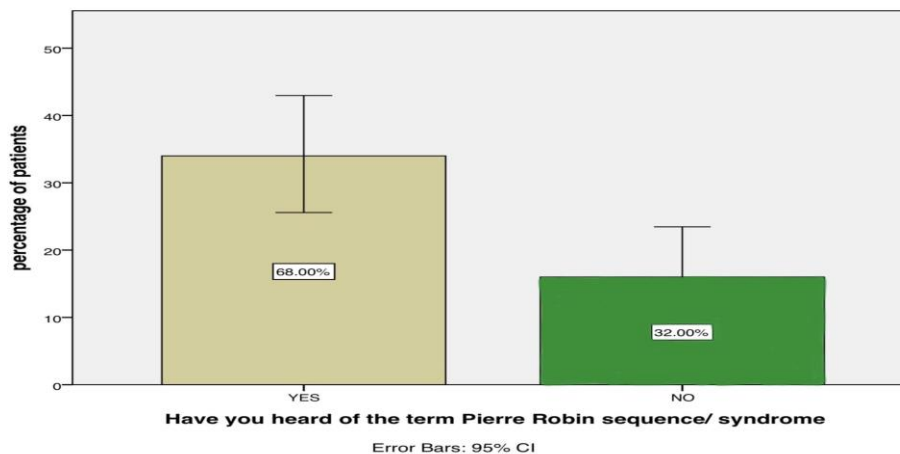


Figure 5-This figure shows a bar graph about pierre robin syndrome plotted against awareness of the participants. The brown colour represents yes, and the green colour represents no. From the graph 68% of the participants are aware of pierre

robin syndrome and 32% of the participants are not aware of stickler syndrome. The chi square test was evaluated for this graph and obtained p value of  $p = 0.184$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

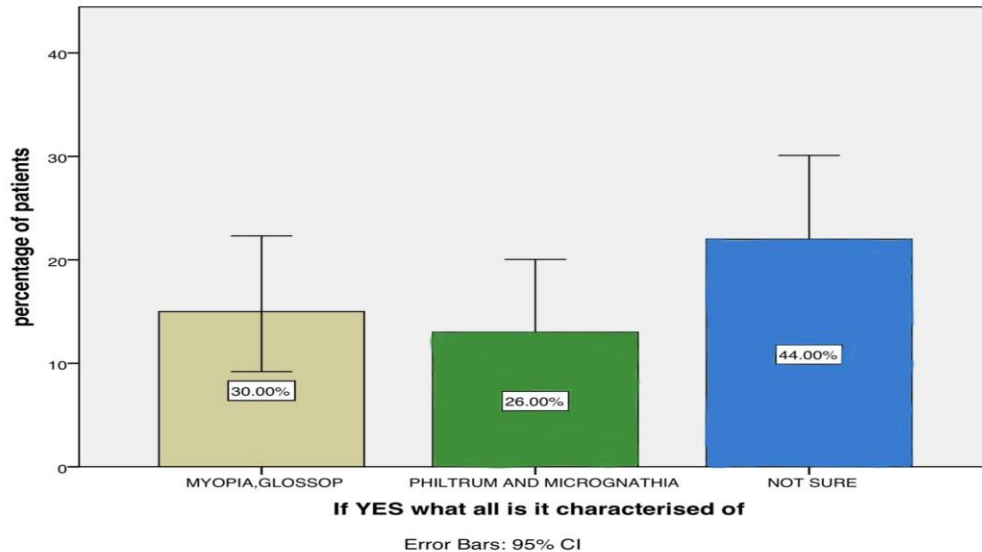


Figure 6 - This figure shows a bar graph about the characteristic feature of pierre robin syndrome against the awareness among the participants. The brown colour represents myopia and glossoptosis, the green colour represents philtrum and micrognathia, the blue colour represents not sure. From the graph 30% of the participants said myopia and glossoptosis are the characteristic features of pierre robin syndrome. 26% of the

participants said philtrum and micrognathia are the characteristic features of pierre robin syndrome. 44% of the participants are not sure about the characteristic feature of pierre robin syndrome. The chi square test was evaluated for this graph and obtained p value of  $p = 0.294$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

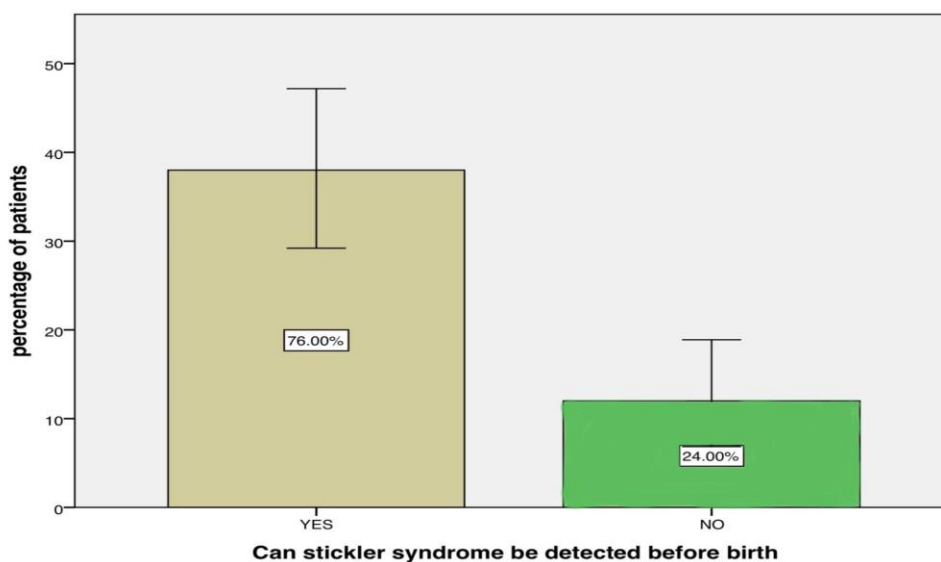


Figure 7 - This figure shows a bar graph about the characteristic feature of pierre robin syndrome against the awareness among the participants. The

brown colour represents yes and green colour represents no. From the graph 76% of the participants said that stickler syndrome can be

detected before birth, and 24% of the participants said that stickler syndrome cannot be detected before birth. The chi square test was evaluated for

this graph and obtained p value of  $p = 0.294$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

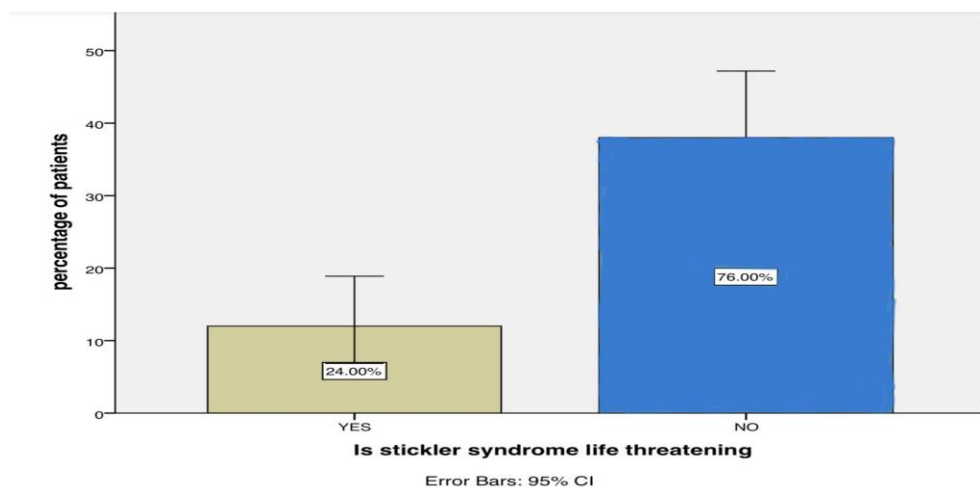


Figure 8 - This figure shows a bar graph as if stickler syndrome is life threatening against the awareness among the participants. The brown colour represents yes, the blue colour represents no. From the graph 24% of the participants said yes and said the stickler syndrome is life

threatening whereas 76% of the participants say that stickler syndrome is not life threatening. The chi square test was evaluated for this graph and obtained p value of  $p = 0.564$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

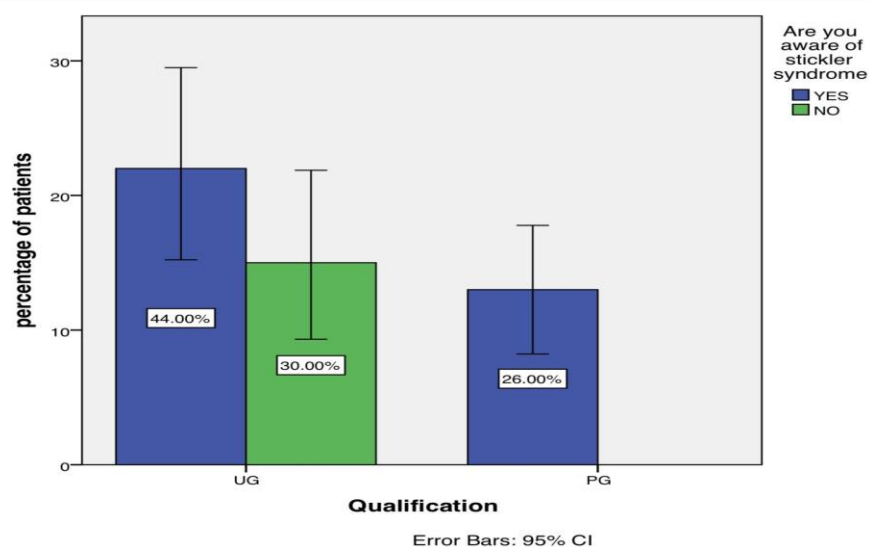


Figure 9- depicts a correlation graph between qualification and the awareness about stickler syndrome among the number of participants. The blue colour represents yes, and the green colour represents No. From the graph it can be seen that 44% of the UG participants are aware of what stickler syndrome is and 30% of the UG students

are not aware of stickler syndrome. Whereas in the PG only 26% of the participants are aware about stickler syndrome. The chi square test was evaluated for this graph and the obtained p value of  $p = 0.367$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

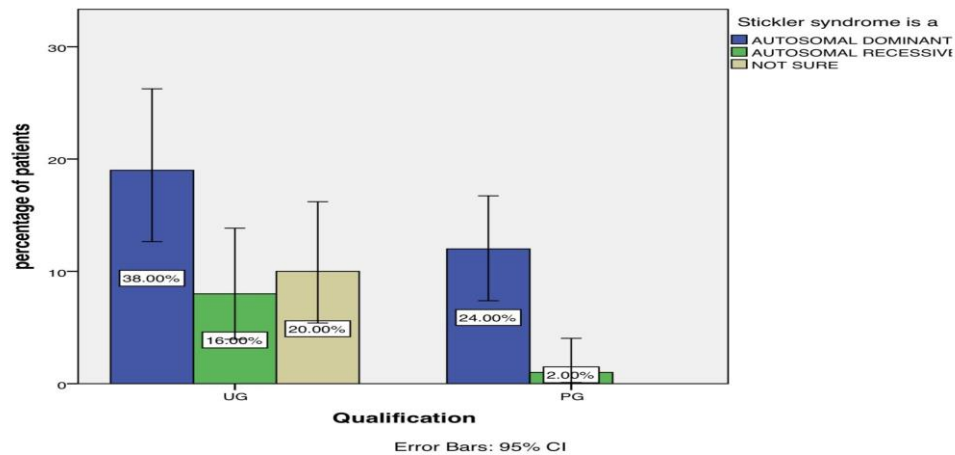


Figure 10- depicts a correlation graph between qualification and the awareness about stickler syndrome if it is autosomal dominant or autosomal recessive or if they are not sure. The blue colour represents autosomal dominant, and the green colour represents autosomal recessive and brown colour represents that they are not sure. From the graph it can be seen that 38% of the UG participants said the stickler syndrome is autosomal dominant, 16% of the UG participants

said stickler syndrome is autosomal recessive, and 20% of them are not sure. From the PG graduate 24% said that it was autosomal dominant, and 2 % of the participants said its autosomal dominant. The chi square test was evaluated for this graph and the obtained p value of  $p = 0.173$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

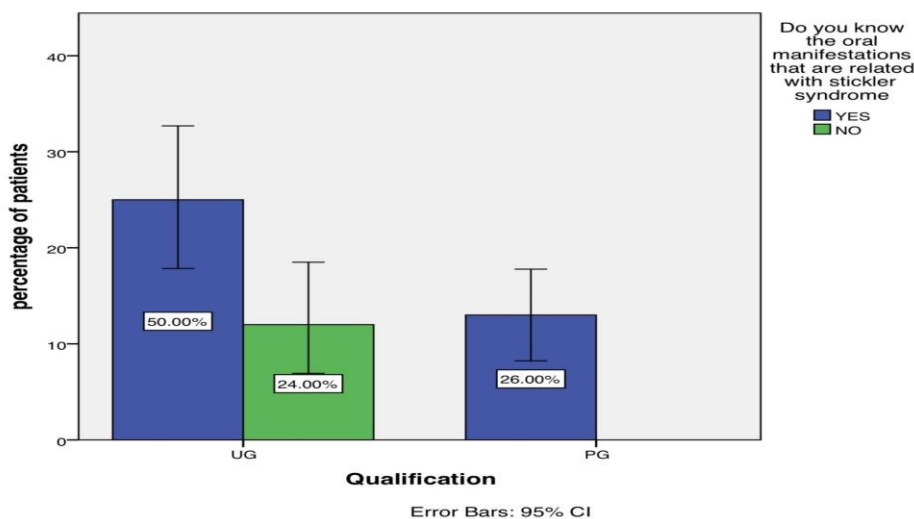


Figure 11-depicts a correlation graph between qualification and if they are aware about oral manifestation of stickler syndrome. Blue colour represents yes, and green colour represents no.

From the graph it can be seen that 50% of the UG participants said the oral manifestations are related with stickler syndrome and 24% of the UG students said the stickler syndrome does not



have any related oral manifestations. 28% of the PG students said that oral manifestations are related with stickler syndrome. The chi square test was evaluated for this graph and the obtained p value of  $p = 0.156$  ( $p > 0.05$ ). Hence the value is statistically insignificant.

## DISCUSSION

A total of 100 students participated in the survey, where 74% of them were UG students and 26% of them were PG students (figure 1). In the survey 69.40% of the UG students were aware about stickler syndrome and 30.60% of the PG students were aware about stickler syndrome (figure 2). Stickler syndrome is a genetic disorder that can cause serious vision, hearing and joint problems. Also known as hereditary progressive arthroophthalmopathy, Stickler syndrome is usually diagnosed during infancy or childhood. 62% of the participants said that stickler syndrome is autosomal dominant and 18% of the participants said its autosomal recessive and 20% of the students were not sure if stickler syndrome is autosomal dominant or recessive (figure 3). Many studies have proved that stickler syndrome is an autosomal dominant multisystem disorder that can affect the eyes, ears, skeleton and joints, and craniofacial (29). Complications may include myopia, cataract, and retinal detachment; hearing loss that is both conductive and sensorineural; midfacial underdevelopment and cleft palate; and mild spondyloepiphyseal dysplasia and/or arthritis. Is there any oral manifestation related with stickler syndrome 76% of the participants said yes and 24% of the students said no (figure 4). Children who have Stickler syndrome often have distinctive facial features — prominent eyes, a small nose with a scooped-out facial appearance and a receding chin. They are often born with an opening in the roof of the mouth (cleft palate). 68% participants know what pierre robin syndrome is and 32% of the participants don't know what pierre robin syndrome is (figure 5).

The Pierre-Robin Syndrome (PRS) is a rare congenital abnormality, with an approximately 1/30,000 estimated rate, characterized by the presence of the combination of mandibular hypoplasia (micrognathia or small jaw), glossoptosis (retrusion of the tongue into the

pharyngeal airway) and, often, a posterior cleft of the secondary palate. It may be an isolated occurrence or part of a more complex syndrome and it is associated with long-term respiratory, nutritional, and developmental difficulties. Stickler syndrome (SS) is a rare autosomal dominant connective tissue disorder estimated to affect approximately 1/7500 newborns. It is diagnosed clinically and, at present, there is no consensus on a minimal clinical diagnostic criterion. The most frequent diagnosis in patients with syndromic Pierre Robin sequence is Stickler syndrome, which may be complicated by congenital high myopia and substantial risk of retinal detachment (1). 30% of the participants said that myopia and glossoptosis are the characteristic of pierre robin syndrome, 26% of the participants said that philtrum and micrognathia are the features of pierre robin syndrome and 44% of the participants are not sure about the characteristics of pierre robin syndrome (figure 6). 24% of the participants said that stickler syndrome is life threatening and 76% of the participants said that it's not life threatening (figure 7). 76% of the participants said that stickler syndrome can be detected before birth and 24% of the participants said that stickler syndrome cannot be detected before birth (figure 8). Among the participants 44% of the UG students are aware about stickler syndrome and 30% of the participants are not aware about the stickler syndrome, 26% of the PG students are aware about stickler syndrome (figure 9). 38% of the UG students said that stickler syndrome is autosomal dominant, 16% of the UG students said it is autosomal recessive, and 20% are not sure. Whereas 24% of the PG students said it was autosomal dominant and 2% said it was autosomal recessive (figure 10). 50% of the UG students said that stickler syndrome is related with oral manifestations and 24% of them said it's not related. 26% of the PG students said it's related with oral manifestations (figure 11).

While Stickler syndrome can sometimes be diagnosed based on your child's medical history and a physical exam, additional tests are needed to determine the severity of the symptoms and help direct treatment decisions. Tests may include: imaging test, eye exams and hearing

test(30) . Genetic testing is available to assist in diagnosis in some cases. Genetic testing can also be used to help in family planning and to determine your risk of passing on the gene to your children when the hereditary pattern is not clear from the family history. Genetic counseling should be provided for affected people. There is no cure for stickler syndrome. We can only prevent the signs and symptoms of stickler syndrome by speech therapy, physical therapy and certain surgeries can also be done.

Most of the dental students know about the term stickler syndrome but they are not sure about the clinical features, diagnosis and treatment for it. This shows that the dental students have a good analysing skill in terms of stickler syndrome but not sure what the condition really means.

## CONCLUSION

Children who have Stickler syndrome often have distinctive facial features — prominent eyes, a small nose with a scooped-out facial appearance and a receding chin. They are often born with an opening in the roof of the mouth (cleft palate). While there is no cure for Stickler syndrome, treatments can help control symptoms and prevent complications. In some cases, surgery may be needed to correct some of the physical abnormalities associated with Stickler syndrome. Jaw surgery. Surgeons can lengthen the lower jaw by breaking the jawbone and implanting a device that will gradually stretch the bone as it heals. Cleft palate repair. Babies born with a hole in the roof of the mouth (cleft palate) typically undergo surgery in which tissue from the roof of the mouth may be stretched to cover the cleft palate. Thus from the study it is clear that most of the dentists have heard of the term pierre robin syndrome but they are not aware about the clinical features, complications and treatment.

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## CONFLICT OF INTEREST :

None to declare.

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