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Awareness On Cornelia De Lange Syndrome Among Dental Students

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Abstract

Introduction:
Cornelia de Lange syndrome is a rare and clinically variable disorder that affects multiple organs. Congenital anomalies include malformations of the upper limbs, gastrointestinal malformation/rotation, pyloric stenosis, diaphragmatic hernia, heart defects and genitourinary malformations.

Aim:
The aim of this study is to observe the awareness level among dental students on Cornelia De Lange syndrome.

Materials and methods:
A cross-sectional questionnaire was created which deals with a check-in awareness level of the dental student population about Cornelia De Lange syndrome. The survey was taken from 500 participants, regardless of their age. The statistical analysis used was SPSS software.

Results:
About 49.33% of the participants say that infants show characteristic features of the syndrome. About 93.33% of the participants say that the origin of the syndrome is congenital. About 69% of participants say that growth delays, intellectual disability and limb abnormalities are noticed in the people suffering from Cornelia De Lange syndrome. About 24.67% of participants think that delayed eruption, dental caries micrognathia are the dental findings observed in people with Cornelia De Lange syndrome.

Conclusion:
More awareness on dental and facial characteristics of Cornelia De Lange syndrome has to be improved, since dental students mostly have a higher chance of encountering patients with this syndrome.

Keywords: Cornelia De Lange syndrome, dental students, facial characteristics

INTRODUCTION:

Cornelia de Lange Syndrome is a rare, genetically heterogeneous disorder that affects multiple organs and systems. The first cases were reported by Dutch anatomists Gerardus and Willem Vrolik in 1849, German physician Brachmann in 1916, followed by Dutch pediatrician Cornelia de Lange in 1933, after whom the syndrome is named[1,2]. CdLS is a clinically variable disorder mainly characterized by psychomotor delay and intellectual disability, distinctive facial features, pre- and postnatal growth retardation, hirsutism, and malformations of especially the upper limbs[3].

The characteristic craniofacial features are pathognomonic for CdLS and lead to the initial diagnosis. In classic CdLS the head is often small (microcephaly), the neck is short and both the anterior and posterior hairline is low[4–7]. The facial features include well-defined and arched eyebrows with synophrys; narrow palpebral fissures; ptosis; long and curly eyelashes; thick, often dysplastic, low set and posteriorly rotated ears; flat midface; short broad nose with a depressed nasal bridge; anteverted nares; long, smooth and prominent philtrum; thin vermillion border of the upper lip; down-turned corners of the mouth; and micrognathia[8–11].

The ear canal is often narrow or stenotic, which predisposes to otitis media and sinusitis. The palate is high and there can be a cleft which is often submucous. Dental anomalies are also observed. Patients with classic CdLS usually have pre- and postnatal growth retardation and a small stature which persists throughout life. Truncal obesity is also seen in some adult patients. Growth can be monitored using growth curves specially developed for CdLS. Psychomotor delay and intellectual disability range from mild learning disabilities to severe impairments.Thus the main aim of the present study is to observe the awareness level of Corenelia De Lange syndrome among dental students.

MATERIAL AND METHOD:

The study is an online survey among the adult population with a sample size of 500 participants. Sampling method was random sampling regardless of the age group. A well structured questionnaire was prepared and circulated using online google forms link to the general population. The purpose of the study was explained to the participants who took the survey, analytics were made using SPSS version 23.0 and to check the association Chi square analysis was done.

RESULTS AND DISCUSSION:

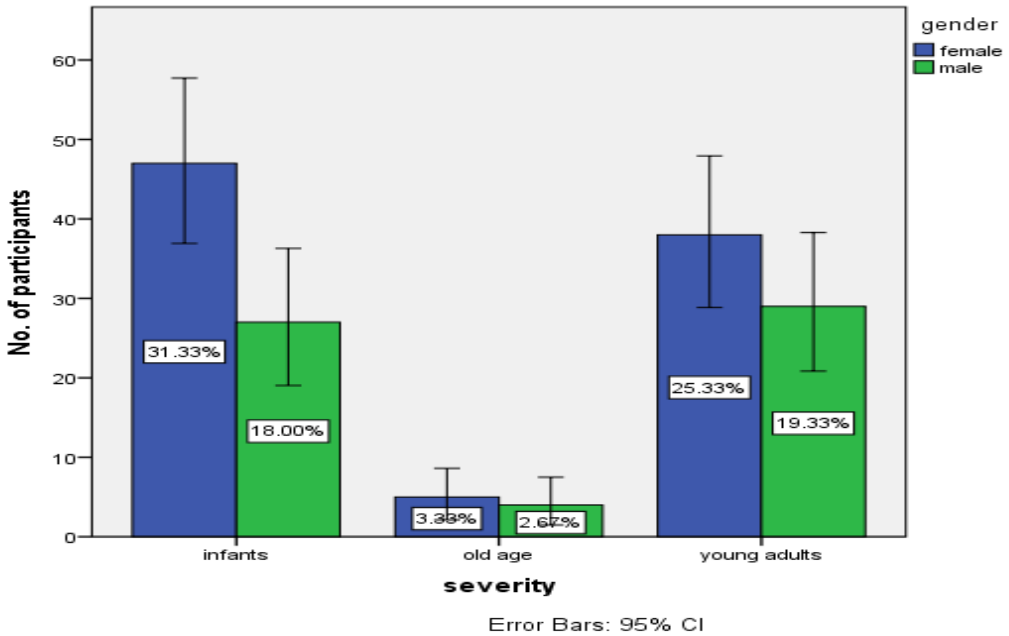

Figure 1: bar graph depicting the correlation of gender of the participants and awareness on severity distribution of people suffering from Cornelia De Lange syndrome. Blue colour denotes female population and blue colour denotes male population of the study. The number of participants are plotted against Y axis and the sawarness on severity of Cornelia De Lange syndrome of the population are plotted under X axis. Chi square test was done and p value obtained is 0.30> 0.05 which is statistically insignificant.

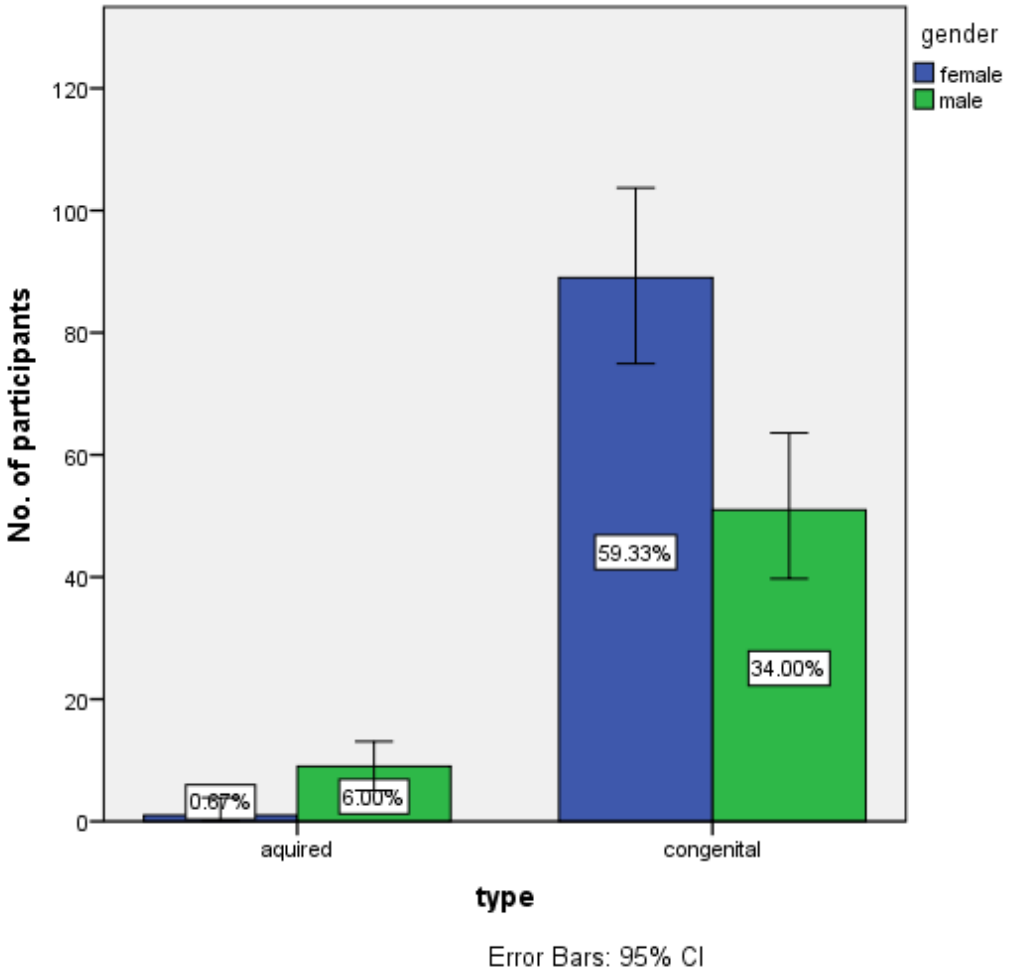

Figure 2: bar graph depicting the correlation of gender of the participants and awareness on type of origin of Cornelia De Lange syndrome. The number of participants are plotted against the Y axis and the awareness on the type of origin of the syndrome of the population are plotted under the X axis. Blue colour denotes female population and green colour denotes male population. Chi square test was done and p value obtained is 0.270> 0.05 which is statistically insignificant.

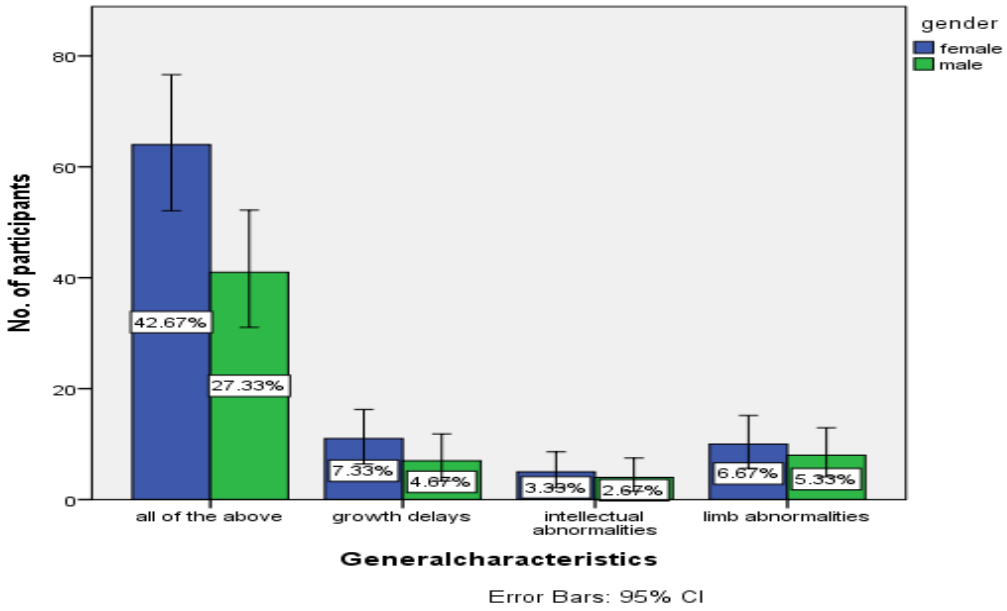

Figure 3: Bar graph depicting the correlation of gender of the participants and awareness on general characteristics of people suffering from Cornelia De Lange syndrome. The number of participants are plotted against Y axis and the awareness level of dental students on general characteristics of people suffering from Cornelia De Lange syndrome are plotted under X axis. Blue colour denotes female and green colour denotes male. Chi square test was done and p value obtained is 0.34> 0.05 which is statistically insignificant.

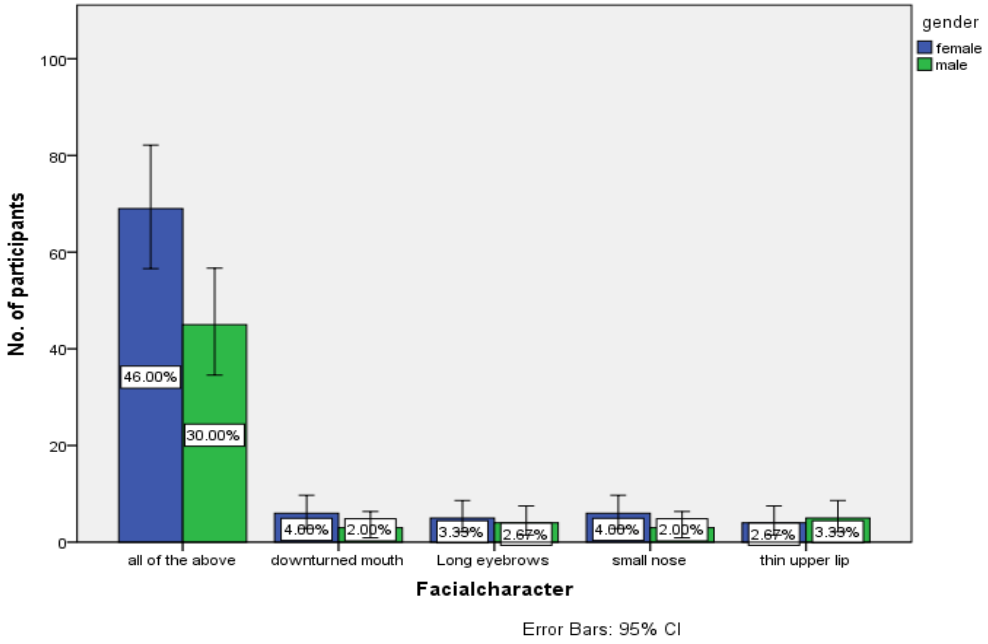

Figure 4: Bar graph depicting the correlation of gender of the participants and awareness on facial characteristics of people suffering from Cornelia De Lange syndrome. The number of participants are plotted against the Y axis and the awareness level of participants on facial characteristics of the syndrome are plotted under the X axis. Blue colour denotes female and green colour denotes male. Chi square test was done and p value obtained is 0.40> 0.05 which is statistically insignificant.

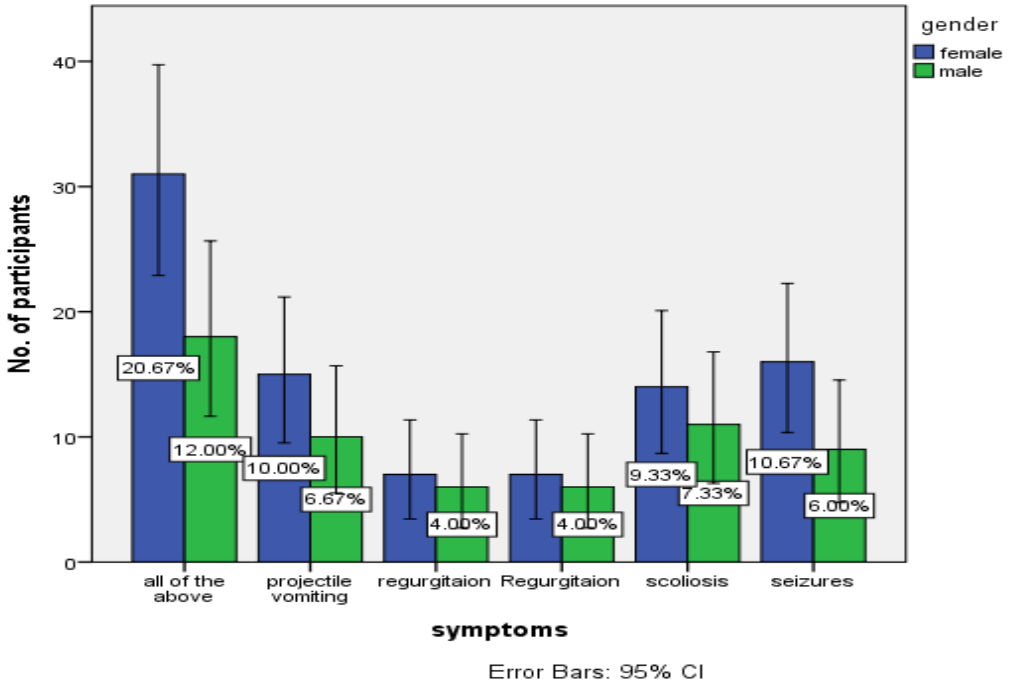

Figure 5: Bar graph depicting the correlation of gender of the participants and awareness on symptoms of people suffering from Cornelia De Lange syndrome. The number of participants are plotted against the Y axis and the awareness level on symptoms of the syndrome among the participants are plotted under the X axis. Blue colour denotes female and green colour denotes male. Chi square test was done and p value obtained is 0.33> 0.05 which is statistically insignificant.

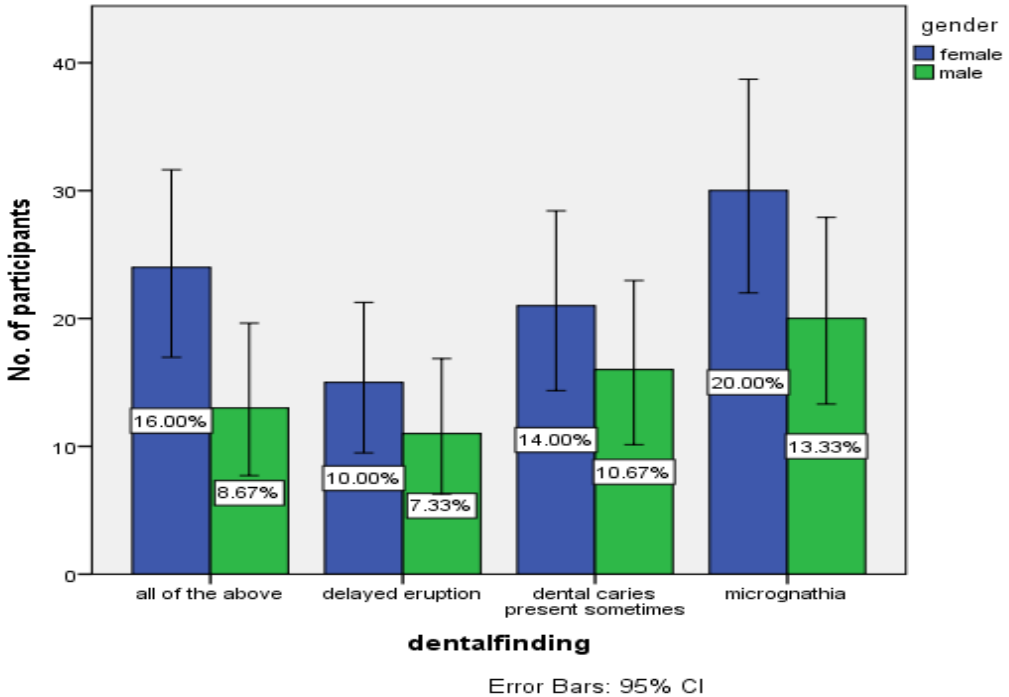

Figure 6: Bar graph depicting the correlation of gender of the participants and awareness on dental findings of people suffering from Cornelia De Lange syndrome. The number of participants are plotted against Y axis and the awareness level on dental findings of the syndrome among the

participants are plotted under X axis. Blue colour denotes female and green colour denotes male. Chi square test was done and p value obtained is 0.26> 0.05 which is statistically insignificant.
From figure 1, it is observed that 31.33% of female and 18% of male participants think that Cornelia De Lange syndrome is mostly in infants. About 3.33% of female and 2.67% of male participants say that the syndrome is associated with old age groups and 25.33% of female and 19.33% of male participants said that the syndrome is mostly associated with young adults. From figure 2, considering the correlation between gender and type of origin of the syndrome, 0.67% of female participants and 6% of male participants think that it is acquired. 59.33% of female and 34% of male participants think that it is congenital in origin. From figure 3, it is observed that considering the general characteristics, 7.33% of female and 4.67% of male participants say that it is growth delays, while it is intellectual abnormalities by 3.33% of female and 2.67% og male participants. Limb abnormalities are the main characteristics of 6.67% of female and 5.33% of male participants, while all of the above are considered as main characteristics of Cornelia De Lange syndrome by 42.67% of females and 27.33% of male participants. From figure 4, it is evident that considering the facial characteristics of the people suffering from Cornelia De Lange syndrome, 4% of females and 2% of male think that a downturned mouth is the main facial characteristics of the syndrome. About 3.33% of female and 2.67% of male participants say that it is long eyebrows while 4% of female and 2% of male participants say that it is a small nose as the facial characteristics of the syndrome. 2.67% of female and 3.33% of male participants think that a thin upper lip is the main facial characteristic noticed in the syndrome. About 46% of female participants and 30% of male participants say that all the above mentioned facial features are observed in the patient suffering from the syndrome. From figure 5, considering the symptoms of the patient reporting hospital suffering from the syndrome, it is observed that projectile vomiting is considered as the main symptom by 10% of female and 6.67% of male participants. Regurgitation is the symptom of the syndrome by 2.22% of female and 4% of male participants. Scoliosis as the main symptom by 9.33% of female 7.33% of male participants. Seizures as the main symptoms by 10.67% of female participants and 6% of male participants. About 20.67% of females and 12% of male think that all the above mentioned symptoms are observed in the patient suffering from Cornelia De Lange syndrome.

Behavioral problems such as hyperactivity, short attention span, attention deficiency and hyperactivity disorder, aggression, defiance, extreme shyness, perseveration, obsessive–compulsive disorders, depression and sleep disturbances have been observed in classic CdLS patients. Many of the behavioral issues are thought to be secondary to frustration from inability to communicate or gastroesophageal reflux disease. Hearing loss is observed in the majority of CdLS patients including both sensorineural and conductive hearing loss. The ear canals are often narrow or stenotic, which predisposes the patients to otits media and sinusitis. The most common ophthalmological findings are high myopia, ptosis, and blepharitis. Nasolacrimal duct obstruction, nystagmus, cataract and glaucoma is observed. GERD is a significant symptom present in over 90% of the patients. Individuals with CdLS and GERD often exhibit “atypical signs' ' as hyperactivity, vomiting and nocturnal agitation. Pyloric stenosis is the most frequent cause of persistent vomiting in the newborn period observed in some of the patients. Other gastroesophageal abnormalities include intestinal malrotation and congenital diaphragmatic hernia.

Mutations in RAD21 (8q24.11) and SMC3 (10q25.2) have been described only in a few patients[12]. The four intragenic RAD21 mutations described to date are: two missense mutations, a single-base pair duplication and an in-frame deletion of exon 13 inherited from a mildly affected mother. Until recently the only identified SMC3 mutation was a 3-bp in-frame deletion in a male patient with mild symptoms. The phenotypic features of the patient included severe microcephaly and atypical facial appearance[13]. Our team has extensive knowledge and research experience that has

translated into high quality publications [14–22],[23–28],[29–35].

CONCLUSION:

From the sblove study, it is evident that knowledge on Cornelia De Lange syndrome is moderate among dental students. More awareness on dental and facial characteristics of Cornelia De Lange syndrome has to be improved, since dental students mostly have a higher chance of encountering patients with this syndrome. Knowledge and awareness about this syndrome helps to diagnose the syndrome at an early stage and for the acute and precise treatment which can be provided to the patient suffering from Cornelia De Lange syndrome.

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CONFLICT OF INTEREST;

Authors declare no potential conflict of interest.

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