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Awareness On Sotos Syndrome Among Dental Students

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Abstract

INTRODUCTION:

Sotos syndrome is a rare genetic disorder which is characterised by a triad of distinctive facial appearance overgrowth in childhood ,learning disabilities and also associated with a wide range of abnormalities. Sotos syndrome also characterizes a unique constellation of signs and symptoms including musculoskeletal findings such as scoliosis ,generalized ligamentous laxity and increased growth rates. According to the recent findings the NSD1 gene mutation was responsible for Sotos syndrome and its genetic disorders with a wide NSD1 alteration.Sotos syndrome is also not life threatening when compared with other syndromes and Sotos syndrome can also be treated and prevented globally among infants and children.

AIM: The aim of this study is to analyse the awareness on Sotos Sotos syndrome among dental students.

MATERIALS AND METHOD:

A questionnaire based survey was conducted among dental students to assess the level of knowledge and awareness about Sotos syndrome. The sample size of this study is 100. The questionnaire consisted of 15 questions. Online study setting was used to collect data. Results were analysed using SPSS software.

RESULTS :

The data was imported to SPSS version 23 and the results were obtained using Chi-square test. Out of which only 36.84% of students were aware of the term Sotos syndrome,40.35% of them said that the term Sotos Syndrome refers to Overgrowth syndrome,77.19% of students said it was mostly infants and children affected,61.40% of students were not aware of the diagnosis,33.33% & 35.09% of them said Sotos Syndrome can be prevented and treated respectively.

CONCLUSION :

From this study we come to know that most dental students are aware of Sotos syndrome but not the causes and effects to which people are affected and also the way it can be treated and prevented.

KEYWORDS**:** sotos syndrome,infants,genetic disorder,defects

INTRODUCTION:

Sotos syndrome is a rare genetic disorder which is characterised by a triad of distinctive facial appearance,overgrowth in childhood ,learning disabilities and also associated with a wide range of abnormalities(1)(2). Sotos syndrome also characterises a unique constellation of signs and symptoms including musculoskeletal findings such as scoliosis (spinal cord abnormalities) ,pes planus,generalised ligamentous laxity and increased growth rates(3). As part of this syndrome ,cervical instability is unreported and rarely presents as a C3 and C4 anterolisthesis(4). According to the recent findings,the NSD1 gene mutation was responsible for Sotos syndrome and its genetic disorders with a wide NSD1 alteration(5)(6). Studies have reported a significant ethnic difference in the frequencies of mutations versus non-mutations in Sotos syndrome with a high percentage of microdeletions(7).Our team has extensive knowledge and research experience that has translate into high quality publications (8–16),(17–22),(23–30) The aim of this study is to analyse the awareness on Sotos Sotos syndrome among dental students.

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RESULTS:

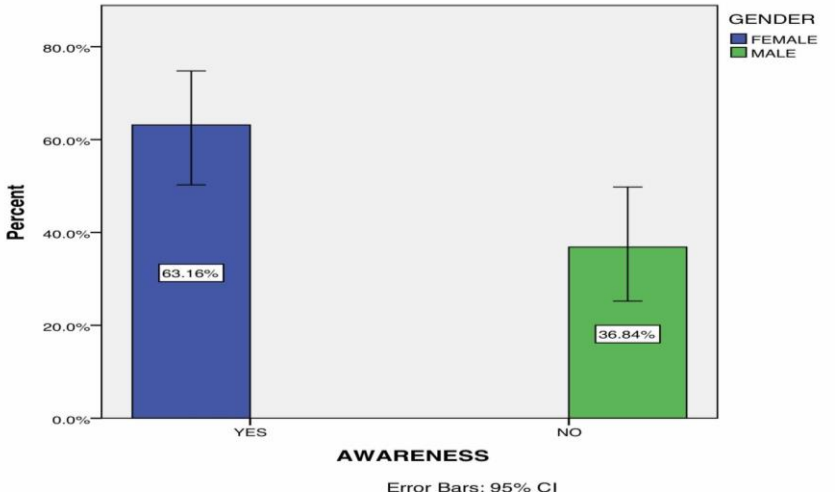

Figure:1 This bar chart shows the correlation between Gender and Awareness on sotos syndrome,where blue colour denotes female and green colour denotes male population.Out of

which only 63.16% people are aware of the term and 36.84% said they were not aware of the term Sotos syndrome.

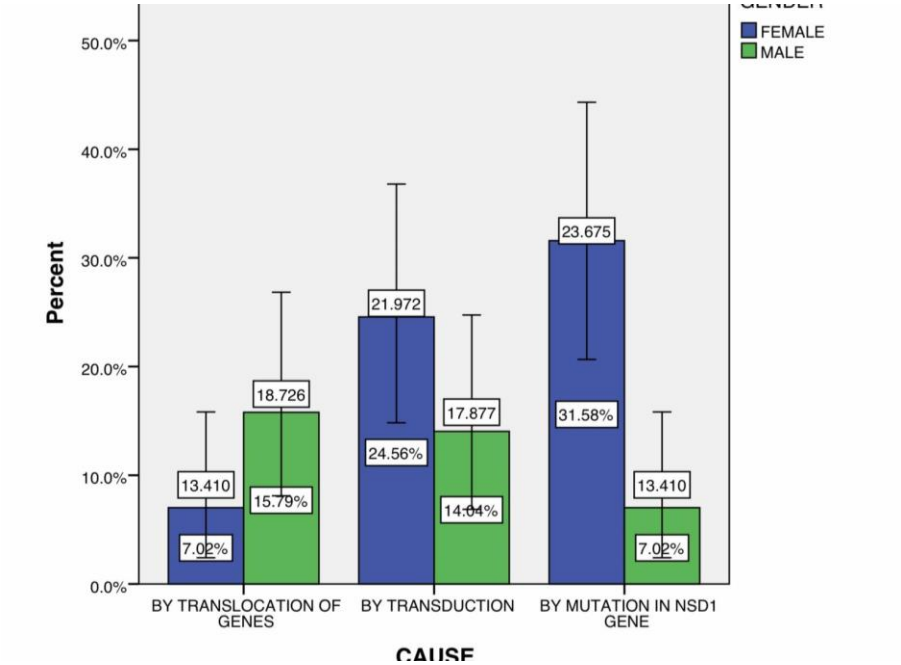

Figure:2 This bar chart shows the correlation between Gender and Cause on sotos syndrome,where blue colour denotes female and green colour denotes male population.Out of which only 22.81% said it was due to mutation of NSD1 gene and 15.79% said it was due to translocation of gene.

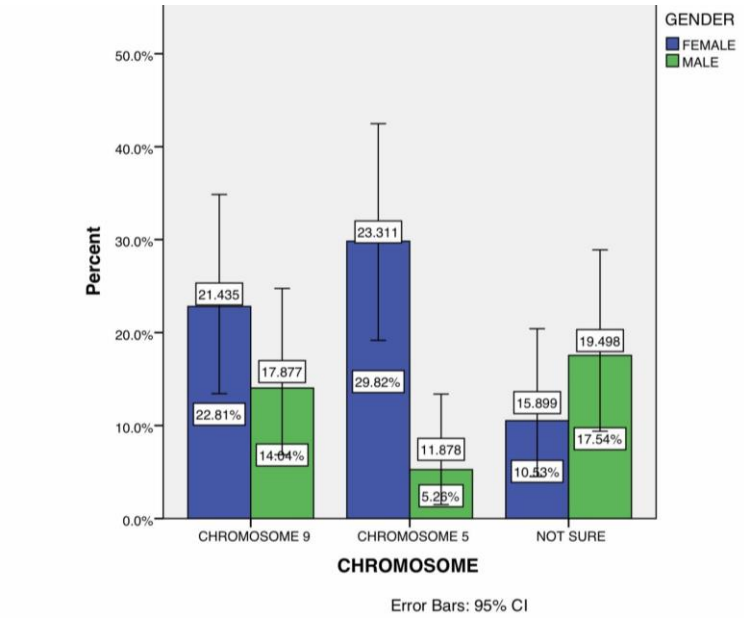

Figure:3 This bar chart shows the correlation between Gender and the Chromosome responsible for sotos syndrome,where blue colour denotes female and green colour denotes male population.Out of which 21.05% said it was due to chromosome 9 , 19.30% said it was due to

chromosome 5 and 14.04% said they were not sure of the chromosome number.

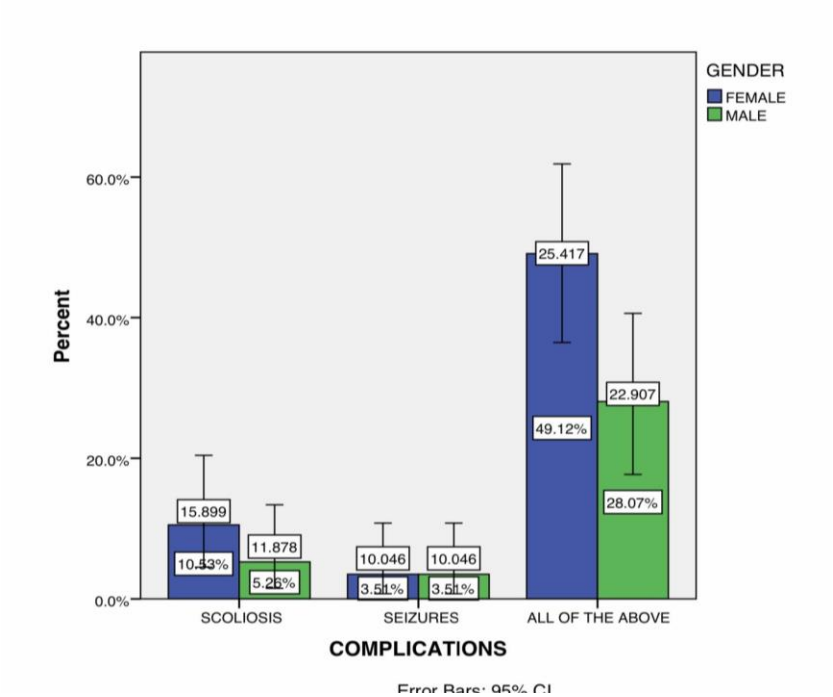

Figure:4 This bar chart shows the correlation between Gender and complications on sotos syndrome,where blue colour denotes female and green colour denotes male population.Out of which only 15.899% said it can be scoliosis and 10.046 % said it can be seizures and 25%.417% saud all the above.

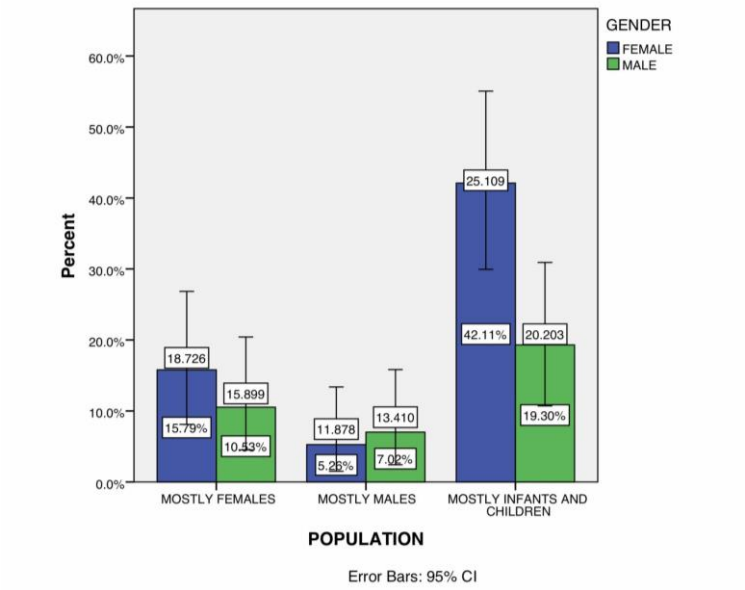

Figure:5 This bar chart shows the correlation between Gender and population affected due to sotos syndrome,where blue colour denotes female and green colour denotes male

population.Out of which 25.10% said it affected mostly infants and children,18.726% said it affected mostly females and 13.51% said it affected mostly males.

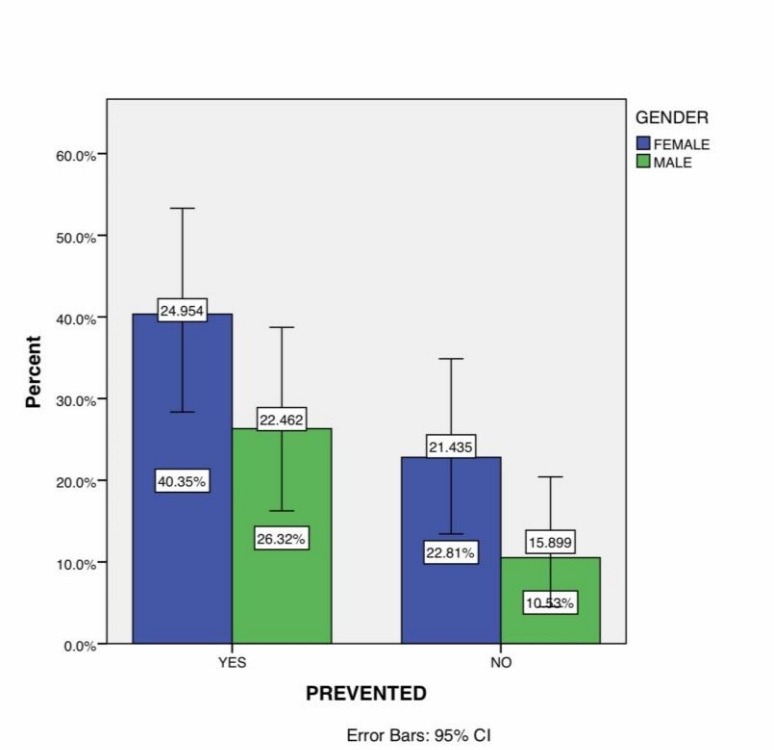

Figure:6 This bar chart shows the correlation between Gender and Prevention of Sotos syndrome,where blue colour denotes female and green colour denotes male population.Out of which 24.954% said it can be prevented and 21.35% said it cannot be prevented.

DISCUSSION:

Molecular basis:

The major cause of Sotos syndrome is halpoinsufficieny of the NSD1 gene at 5q35 that plays a major role in growth and brain development in humans(31). This Sotos is due to an NSD1 point mutation is a single gene defect, occasionally with an autosomal dominant mode of inheritance.(31)Apart from this Cerebral gigantism, hypotonia and joint hyperextensibility are characteristic features of this syndrome(32). Mutations in NSD1 are responsible for Sotos syndrome, but are not a frequent finding in other overgrowth phenotypes(2,33).

Genetic syndromes:

In Sotos Syndrome, overgrowth syndromes comprises a diverse group of conditions with unique clinical and molecular genetic features(34). The best correlation between clinical assessment and molecular results was Sotos facial gestalt in conjunction with overgrowth, macrocephaly and developmental delays(2,34).

Due to this haploinsufficiency of the NSD1 gene on chromosome 5q35.2-35.3 in 90% of people: Sotos syndrome 1 recently heterozygous mutations in the NFIX gene on chromosome 19p13.3 where identified in few children with Sotos syndrome phenotype :Sotos syndrome 2(35).The disease is said to be not uncommon and can be missed easily in a clinical setting, if the clinician is not so vigilant and notices the patient. Thus, Sotos syndrome is not a rare disorder and can be diagnosed among children with global developmental delay. Sotos syndrome is also not life threatening when compared with other

syndromes and Sotos syndrome can also be treated and prevented globally among infants and children.

In this study , we assessed the level of awareness about Sotos syndrome among dental students. In this study, 100 participants are involved out of which 90% of them are undergraduates and 10% of them are postgraduates.From this study, we come to know that only 29.82%of the the participants are familiar of this term Sotos syndrome and and the remaining 24.56% are not familiar with the term Sotos Syndrome(**Figure:1**). The term sotos syndrome can be due to defects in facial muscle28.07%, 21.05% due to overgrowth syndrome and 7.02% said it was due to affected intelligence. On assessing what Sotos syndrome means,55.8% said that it is a rare genetic disorder,23.1% said it is a rare acquired disorder and 21.2% had no idea about Sotos syndrome . According to (**Figure:2**) 22.81% of people were aware that Sotos syndrome was caused by mutation in the NSD1 gene,whereas the remaining 22.81% and 15.79% said Sotos syndrome is caused by transduction and by translocation of genes respectively. The participants were also asked about the chromosome number affecting Sotos syndrome out of which 19.30% of people said chromosome 5,remaining 21.05% said it can be chromosome 9 (**Figure:3**). The participants were asked about the features of Sotos syndrome for which 59.6% of people said All the above and remaining 19.2% and 17.3% said it can be distinctive facial appearance and overgrowth in childhood respectively. 57.4% of participants said Sotos Syndrome can be life threatening whereas on the other hand 42.6%of participants said Sotos syndrome cannot be life threatening.On assessing the complications of Sotos syndrome 38.60% said it causes scoliosis and 24.56% said it causes seizures (**Figure:4**).The population group affected by Sotos syndrome was 47.37% said it was mostly infants and children ,12.28% said it was females and 3.51% said it was males mostly affected(**Figure:5**) The diagnostic method for Sotos syndrome is "Genetic test" said by 33.3%, "MRI" said by 16.7% and "Biopsy"said by14.8%. Participants were asked about the treatment for Sotos syndrome for which 63% said "yes" and 37% said that they "no". Participants were also asked whether Sotos syndrome can be prevented for which 38.60% said Yes and the remaining 17.54% said No (**Figure:6**).

Sotos Syndrome is an overgrowth condition characterised by cranial features including excessive growth during ,macrocephaly,distinctive facial gestalt and various degrees of learning.According to previous researchers, Sotos Syndrome are due to mutations and depletion of the NSD1 gene which are responsible for more than 75% of cases.Many articles have shown that during the neonatal period ,therapies are mostly symptomatic,including phototherapy in case of jaundice ,treatment of the feeding difficulties and the gastroesophageal reflux,and detection and treatment of hypoglycaemia.As Sotos syndrome affects most of the infants and children, general pediatric follow - up is important during the first years of life to allow detection and management of clinical complications such as scoliosis and febrile seizures.

An adequate psychological and educational program with speech therapy and motor stimulation plays an important role in the global development of the patients.Sotos syndrome can be prevented and treated to a greater extent if it is diagnosed at its early stages of life .

CONCLUSION:

From this study we come to know that most dental students are aware of Sotos syndrome but not the causes and effects to which people are affected and also the way it can be treated and prevented.

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