

## A study on morphological features in relation with co-morbidities of down 's syndrome

Dr. M. Swarna Madhuri<sup>1\*</sup>, Dr. Saikiran Pasupula<sup>2</sup>, Dr. P. Savithri<sup>3</sup>

### ABSTRACT

**Background:** Down syndrome (DS) is the most commonly identified genetic form of mental retardation and the leading cause of specific birth defects and medical conditions. DS is a multi-system disorder with varied clinical features that affects the individual physically, medically and psychologically. **Aims and Objectives:** 1. To study the morphological features of DS and their co morbidities. 2.To study the relation between morphology and co morbidities of DS. **Materials and methods:** This were an observational study done at special school for downs syndrome (n= 60). After prior consent socio demographic data was taken using semi structured proforma and morphological data was noted by general examination and the co-morbidities are noted with the clinical reports available with them. **Discussion:** Morphological features in the current study are varied and supports the earlier literature that DS is a multi-system disorder that affects the individual physically, medically and psychologically. **Results:** Individuals showed diversified clinical features .Results revealed that males are more than females and among the associated clinical features majority of the individuals have flat nose and least number of individuals have nystagmus. **Conclusion:** DS is a genetic anomaly with considerable medical co morbidities. Early identification of co morbidities with proper health education to the parents helps in preventing the major harmful effects and promotes better healthier life.

**Keywords:** Down syndrome, Trisomy 21, Special children, Genetic disorders

**D**own syndrome (DS) is the most commonly identified genetic form of mental retardation and the disorder is the leading cause of specific congenital disabilities and medical conditions. DS or trisomy 21 is a genetic disorder, which is due to the presence of 47 chromosomes instead of 46, with an extra copy of chromosome 21.<sup>[1]</sup> The additional copy of chromosome 21, which may either be full or partial, depending on the variant, causes the abnormality and associated structural and functional anomalies of the bodily systems. <sup>[2]</sup> Down's syndrome is a multi-system disorder with varied clinical features that affect the individual physically, medically and psychologically. Down's syndrome is associated with

<sup>1</sup>Post graduate, Department of Anatomy, Guntur medical college, Guntur, India

<sup>2</sup> Assistant Professor, Department of Psychiatry, Katuri Medical College, Guntur, India

<sup>3</sup> Professor and HOD, Department of Anatomy, Guntur medical college, Guntur, India

\*Responding Author

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much co-morbidity, and the life expectancy of downs can be increased with early identification and management of co-morbidities.

### *Aims and Objectives*

1. To study the morphological features of DS and their co-morbidities.
2. To study the relationship between morphology and co-morbidities of DS.

## **METHODOLOGY**

This study was an observational study done at special school for downs syndrome (n= 60). After prior consent from the parents and school authorities, socio-demographic data was taken using semi-structured proforma, and morphological data were noted by general examination, and the diagnosis of downs was confirmed with the help of karyotyping. Co morbidities were documented with the clinical reports available with them. Statistics was done with the help of EPI INFO software and results were tabulated using M.S.Excel software.

## **RESULTS**

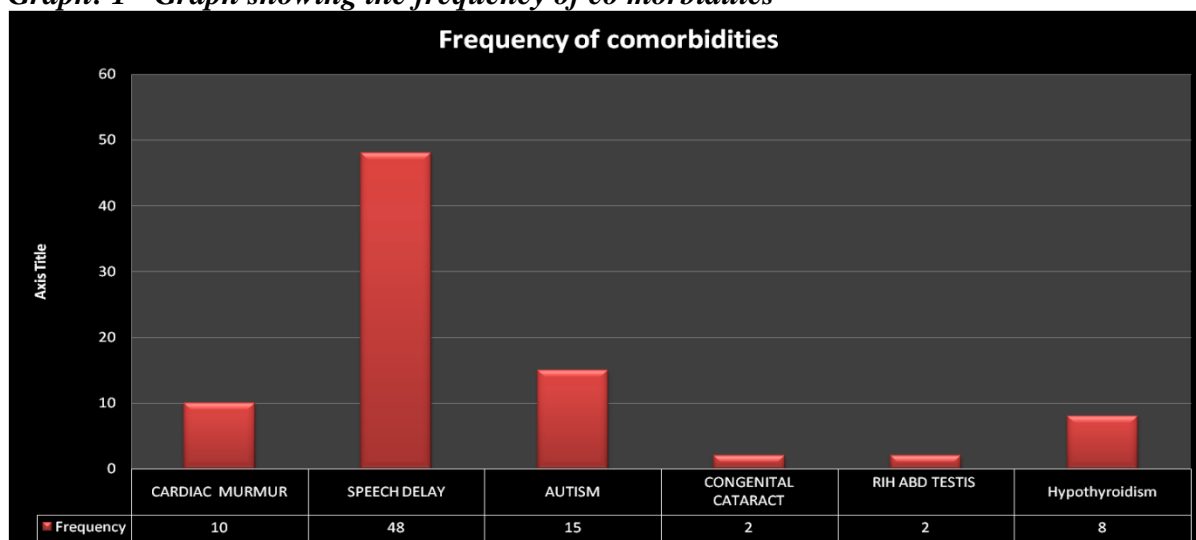
*Table No. 1: Socio-demographic data*

<i>VARIABLE</i>	<i>FREQUENCY</i>
<i>Sex</i>	
<i>Female</i>	<i>18</i>
<i>Male</i>	<i>42</i>
<i>BIRTH ORDER</i>	
<i>First</i>	<i>32</i>
<i>Second</i>	<i>26</i>
<i>Third</i>	<i>2</i>
<i>CONSANGUINITY</i>	
<i>No</i>	<i>54</i>
<i>Yes</i>	<i>6</i>
<i>MATERNAL AGE</i>	
<i>18-20</i>	<i>8</i>
<i>20-30</i>	<i>50</i>
<i>More than 30</i>	<i>2</i>

*Table 2: Frequency of symptoms among the individuals who have Down 's syndrome*

<i>PHENOMENOLOGY</i>	<i>FREQUENCY</i>
<i>Hypertelorism</i>	<i>48</i>
<i>Epicanthal</i>	<i>50</i>
<i>Conical teeth</i>	<i>34</i>
<i>Delayed teeth</i>	<i>2</i>
<i>Flat nasal bridge</i>	<i>48</i>
<i>High arched palate</i>	<i>50</i>
<i>Protruded tongue</i>	<i>16</i>
<i>Nystagmus</i>	<i>16</i>
<i>Squint</i>	<i>2</i>
<i>Brush field spots</i>	<i>16</i>
<i>Short neck</i>	<i>34</i>
<i>Clinodactyly</i>	<i>34</i>
<i>Simian crease</i>	<i>32</i>
<i>Sandal gap</i>	<i>46</i>

Graph: 1 - Graph showing the frequency of co morbidities



## DISCUSSION

The current study advances the earlier studies on downs in studying the phenomenology of downs and associated co-morbidities. Previous studies on downs show that prevalence of downs among males and females is the same and male to female ratio was approximately one. This result is contrary to the finding of the current study that males are more in number. In the present study mean age of the mothers is 28.5 years, and most of the mothers are in the age group of 20-30 years which can be explained by the fact of higher prevalence of reproduction at a younger age. Upper mean age supports the statement done by previous studies that advanced maternal age increases the risk of down's syndrome [3]. Among the co-morbidities, most of the children are suffering from a speech delay. Among them, we found fifteen individuals with autism [4]. Earlier studies found that cardiac murmur was seen among 40-50% of the downs individuals, but in our research, there are only ten children which accounts for only sixteen per cent of the total sample. [5] Hypothyroidism is another common co-morbidity of down's syndrome. It affects 15 - 20% of Down syndrome, and in the current study, we found eight children with hypothyroidism. [6] Two children are with RIH with abdominal testis and congenital cataracts, respectively, which can be an incidental finding among downs.

### Limitation of the study

As the research was done at a single centre, results cannot be generalized. More reviews at the multi centric level are needed for extrapolation of results.

## CONCLUSION

DS is a genetic anomaly with considerable medical co-morbidities. Early identification of co-morbidities with proper health education to the parents helps in preventing the significant harmful effects and promotes a better, healthier life. We need many numbers of similar studies for better validation of results.

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### **Conflict of Interest**

The author declared no conflict of interest.

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