Original Research Article

# Clinical profile of children with down syndrome with special reference to thyroid hormone levels

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## Abstract

**Background:** Down syndrome (DS) results from trisomy of chromosome 21. Down syndrome produces a typical phenotype that includes a characteristic mongoloid facies, mental retardation and hypotonia, and is often associated with developmental anomalies. Children with Down syndrome are at an increased risk of developing hypothyroidism. **Objective:** To study the clinical profile of children with Down syndrome with special reference to thyroid hormone levels. **Methodology:** This study was conducted from Mar 2016–Feb 2018 (2 years) on 35 children with Down syndrome attending the Pediatric Outpatient department (OPD), GEMS and Hospital, Ragolu, Srikakulam and were subjected to thyroid function tests (Free T4 and TSH) at the time of recruitment into the study. Age and sex matched children attending the OPD for well-baby checkup and immunization were taken as controls. **Results:** In the present study, majority (85.7%) of babies with Down syndrome were born to mothers aged less than 35 years. Present study had a 34.3% frequency of congenital heart disease, with VSD being most common type (33%). In the present study, 5 patients (14.3%) were found to have thyroid dysfunction. **Conclusion:** Diagnosis of Down syndrome is done by characteristic phenotype.14.3% of children had hypothyroidism in our study.
**Key Word:** Down's syndrome, Serum TSH and Free T4, Hypothyroidism

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## INTRODUCTION

Down syndrome produces a typical phenotype that includes a characteristic mongoloid facies, mental retardation and hypotonia, and is often associated with developmental anomalies[1]. It is the most common chromosomal disorder with frequency of 1 in 800 to 1 in 1000. Although the probability increases with maternal age, 80% of children with Down syndrome are born to women under the age of 35.[2]Children with Down syndrome have a shorter life expectancy primarily because of infection due to impaired immune response. Both sexes are affected equally. The diagnosis of Down syndrome at birth is based on the presence of a constellation of physical features and is confirmed by genetic karyotyping. Clinical criteria used to diagnose Down's syndrome are: Hall's Ten Signs of Down Syndrome in Newborns: [3] Poor Moro reflex, Hypotonia, Flat facial profile, Upward-slanting palpebral fissures, Morphologically simple, small round ears, Redundant loose neck skin, Single palmar crease, Hyper extensible large joints, Pelvic dysplasia, Short 5$^{th}$ digit with clinodactyly Oster (1953) has listed the following ten cardinal signs in childhood: [4] Flat facial profile, Oblique palpebral fissures, Flat nasal bridge, High archedpalate, Small teeth, Flatocciput, Small or absent lobes of theears, Short broad hands, Short fifth finger, Hyper extensibility

**Congenital heart disease:** Congenital heart defects are present in 40 to 60% of children with Down syndrome.[5] The most common congenital heart defects are endocardial cushion defect (60%) ventricular septal defect (32%), secundum atrial septal defect (10%), tetralogy of fallot (6%) and isolated patent ductus arterious (4%)[6]

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**Down syndrome and thyroid:** Children with Down syndrome are at an increased risk of developing hypothyroidism (25-30%).[7] In fact, an excess of TSH with normal levels of T3 and T4 appear to be frequently associated with Down syndrome[8]. The following reasons have been postulated for thyroid dysfunction in Down syndrome children like it was a kind of resistance syndrome, possibility of a less active TSH and an increased rate of degradation of T4 in the periphery, and the body needs to secrete increased amounts of T4 to maintain a homeostasis.[9,10]

**Table 1:** clinical features of hypothyroidism

| Congenital | Acquired |
|---|---|
| Open Posterior Frontanel | Growth Retardation |
| Umblical Hernia | Delayed Skeletal Maturation |
| Edematous Facies | Delayed Dental Development |
| Constipation | Delayed Puberty |
| Pallor | Myopathy and Pseudohypertrophy |
| Hypothermia | Enlarged Sella |
| Large Tongue | Pseudotumor Cerebri |
| Rough Dry Skin | |
| Hypotonia | |
| Large Abdomen | |

Estimation of Free Thyroid hormone is superior to total levels in the diagnosis of hypothyroidism. Low FT4 and TSH levels suggest central hypothyroidism while high TSH levels indicate primary hyperthyroidism. Persistent elevation of TSH in the presence of normal FT4 suggests subclinical hypothyroidism.

## MATERIALS AND METHODS

Our Study Design was Descriptive study with the Study Period from March 2016 to February 2018 (2 years). Total 35 children with Down syndrome attending the Pediatric OPD, GEMS and Hospital, Ragolu, Srikakulam for well-baby checkup and immunization were included for the study. With the age group less than 15years.

## METHODOLOGY

All children with Down syndrome were enrolled with the following details: Age at diagnosis, age at recruitment into the study, maternal age, and clinical features as per the proforma. All children with Down syndrome were subjected to thyroid function tests (Free T4 and TSH) at the time of recruitment into the study. Age and sex matched children attending the outpatient for well-baby checkup and immunization were taken as controls.

## OBSERVATIONS

A) **Age-Sex Distribution**

**Table 2:** Age and Sex Distribution Of Cases

| Age of patients | Total | | Male | | Female | |
|---|---|---|---|---|---|---|
| | Number of patients | % | Number of patients | % | Number of patients | % |
| **<6months** | 14 | 40 | 07 | 20 | 07 | 20 |
| **6 months- 1 year** | 05 | 14.3 | 04 | 11.4 | 01 | 3 |
| **1-5 years** | 10 | 28.6 | 04 | 11.4 | 06 | 17.1 |
| **5-10 years** | 03 | 8.5 | 03 | 8.6 | 0 | 0 |
| **10-15 years** | 03 | 8.5 | 2 | 5.7 | 1 | 3 |
| **TOTAL** | 35 | 100 | 20 | 100 | 15 | 100 |

In this study, 35 patients with Down syndrome were studied of whom 20 (57.1%) were boys and 15 (42.8%) were girls. The male and female ratio is **1.3: 1**. Majority (40%) of the children were less than 6 months of age followed by 28.6% of the children between 1-5 years of age.

**B) Maternal Age versus Down syndrome**

Of the 35 children studied, 2 (5.7%) mothers were aged less than 20 years at the time of conception age, 9 (25.7%) mothers were between 20-24 years,12 (34.3%) of the mothers were between 25-29 yrs, 7 (20%) of the mothers were between 30-34 years age group, 4(11.4%) of the mothers were between 35-39yrs, 1 (2.9%) of the mothers were between 40-45yrs.

**Table 3:** Maternal Age Versus Down Syndrome

| Age of mother (in years) | Number of children | % |
|---|---|---|
| <20 | 02 | 5.7 |
| 20-24 | 09 | 25.7 |
| 25-29 | 12 | 34.3 |
| 30-34 | 07 | 20 |
| 35-39 | 04 | 11.4 |
| 40-45 | 01 | 2.9 |
| TOTAL | 35 | 100 |

**C) Frequency of Congenital Heart Disease in This Study**

Total of 12 children were found to have heart disease. Most common congenital heart disease detected was VSD.

**Table 4:** Congenital Heart Disease

| Congenital Heart Disease | TOTAL |
|---|---|
| ASD | 2 |
| VSD | 4 |
| PDA | 2 |
| TOF | 1 |
| AVSD | 1 |
| COMBINATION | 2 |

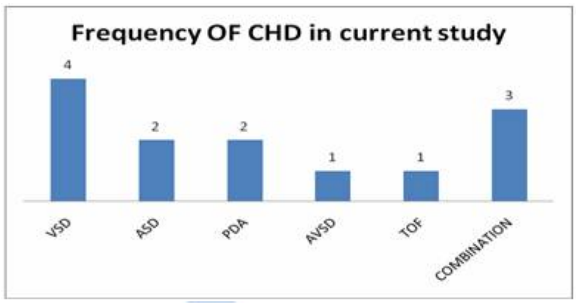


**Figure 1:**

**D) Anthropometry**

Among the 35 children, 11 (31.4%) children had short stature and 11 children had low weight for age.

**Table 5:** Height And Weight

| | Normal | Abnormal |
|---|---|---|
| **Height** | 24 | 11 (Short stature) |
| **Weight** | 24 | 11 (low weight) |

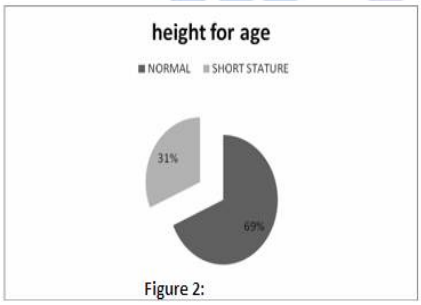


Figure 2:

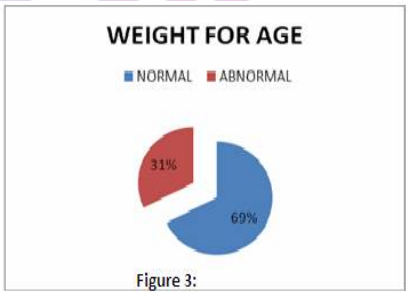


Figure 3:

**Table 6:** Head Circumference

| Head circumference | N= 27 |
|---|---|
| Normal | 15 |
| Microcephaly | 12 |

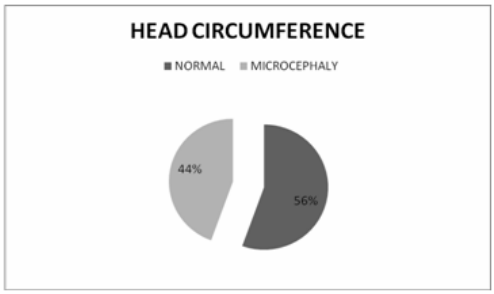


**Figure 4:**

Among 27 children under the age 5 years, 12 children had microcephaly.

**E) hypothyroidism in down syndrome:**

Of the 35 children with Down syndrome, 30 children had normal free T4 and TSH, 2 children had normal free T4 and high TSH and 3 children had low free T4 and high TSH

**Table 7:** Hypothyroidism In Down Syndrome

| Thyroid profile | Total | Females | MALES |
|---|---|---|---|
| Normal | 30 | 13 | 17 |
| Subclinical hypothyroidism | 2 | 0 | 2 |
| Hypothyroidism | 3 | 1 | 2 |
| Total | 35 | 14 | 21 |

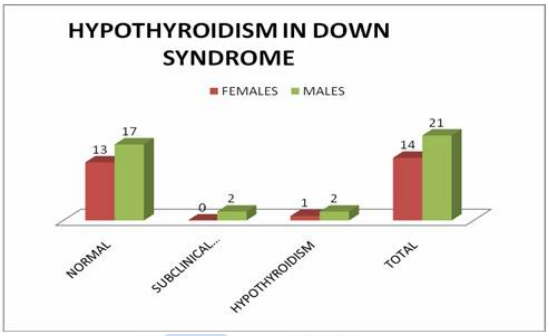


**Figure 5:**

## DISCUSSION

In the present study, 35 children with Down syndrome were studied, of which 20 (51.7%) were males and 15 were females (42.9%). Majority of the children (54.3%) were under 1 year followed by 1- 5 years age group (28.6%) which were similar to studies conducted by Amir Muhamed *et al* from Pakistan[11], by Kava *et al* from India[12] and Azman *et al* from Malaysia. [13] Advanced maternal age is an important risk factor due to chromosome 21 malsegregation. However in the present study, majority (85.7%) of babies with Down syndrome were born to mothers less than 35 years, which is similar to studies conducted by Malini *et al*[14] and Kava *et al*[12] which may be due to genetic predisposition to chromosomal malsegregation. Clinical diagnosis of Down syndrome was suspected in 28 (80%) patients at presentation. The knowledge of clinical manifestations of DS by clinicians is important to make an early postnatal diagnosis possible, since prenatal diagnosis is not frequently performed. Present study had a 34.3% frequency of congenital heart disease. This frequency was similar to the study conducted by Ahmed *et al*[15]where the incidence of CHD was 34.9%. The most common type of CHD in this study was VSD (33.3%). In the present study, 5 patients (14.3%) were found to have thyroid dysfunction. Out of these 5 children, 4 were males and 1 was female child. Of the 4 males, 2 were having sub clinical hypothyroidism and 2 were overt hypothyroidism.

## CONCLUSION

Diagnosis of Down syndrome is done by characteristic phenotype. The frequency of hypothyroidism in children with Down syndrome is 14.3% in our study. Congenital heart disease was present in 34.3% of cases, with ventricular septal defect being the most common type of cardiac defect.

## REFERENCES

1. "CDC Birth Defects, Down Syndrome NCBDDD". Cdc.gov.2013-11-06.
2. Estimate from" National Down Syndrome Center". Retrieved2006-04-21.
3. Hindley, D. (2002). "Diagnosis of Down syndrome in neonates". Archives of Diseasein Childhood-Fetal and Neonatal Edition 87 (3):220F.Oster J,MikkelssenM, NielsenA. Mortality and life table in Down syndrome.ActaPediatrScand1975, 64:322-326.
4. Korenberg J, Kurnit D. Molecular and stochastic basis of congenital heart defects in Down syndrome. In: Marino B, Pueschel SM, eds. Heart disease in persons with Down syndrome. Baltimore: Brookes,1996:21-38
5. Marino B. Patterns of congenital heart disease and associated cardiac anomalies in children with Down syndrome. In: Marino B, Pueschel SM, eds. Heart disease in persons withDown syndrome. Baltimore: Brookes, 1996:133-40.
6. Fabris N, Mocchegiani E, Amadio L, Zannotti M, Licastro F, Franceschi C (1984) Thymic Hormone Deficiency in Normal Ageing and Down Syndrome: is there a Primary Failure of the Thymus? The Lancet May 5;983-986
7. B Bjorksten,O Back, KH Gustavson, G Hallmans, B Hagglof, A Tarnvik; Zinc and immune function in

Down's syndrome . Acta Paediatrica Scandinavica, 69 (1980), pp.183–187
8. Prasher VP. Down syndrome and thyroid disorders: a review. Downs Syndr Res Pract1999; 6:25-42.
9. Pueschel, SM, Pezzullo, JC (1985): Thyroid dysfunction in Down syndrome. Am J Dis. Child139:636–639.
10. Amir Mohammad, Inayatullah Khan, Mohammad Qasim Khan. Gomal Journal of Medical Sciences January-June 2012, Vol. 10, No. 1; 96 -99.92.
11. KavaMP,TulluMS,MuranjanMN,GirishaKM.Downsyndrome:clinical profile from India. Arch Med Res. 2004; 35: 31-5.
12. Azman BZ, Ankathil R, Siti Mariam I, Suhaida MA, Norhashimah M, Tarmizi AB, *et al*. Cytogenetic and clinical profile of Down syndrome in Northeast Malaysia Singapore Med J 2007; 48: 550-4.
13. Suttur S. Malini, Nallur B. Possible risk factors for Down syndrome and sex chromosomal aneuploidy in Mysore, South India. Indian J Hum Genet. 2007 Sep-Dec; 13(3): 102–108. doi:10.4103/0971-6866.38984
14. Ahmed I, Ghafoor T, Samore NA, Chattha MN. Down syndrome: clinical and cytogenetic analysis. J Coll Physicians Surg Pak.2005; 15: 426-9.

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