

# Down syndrome in tribal population in India: A field observation

Ram Lakhan, Madhavaram Thomas Kishore<sup>1</sup>

Department of Epidemiology, Jackson State University, Jackson, MS, USA, <sup>1</sup>Department of Clinical Psychology, National Institute of Mental Health and Neurosciences, Bengaluru, Karnataka, India

## ABSTRACT

**Context:** Down syndrome (DS) is a prevalent genetic disorder in intellectual disability (ID) in India. Its prevalence in tribal population is not known. **Aims:** The study aimed to understand the profile of DS in a tribal population with an objective of finding the prevalence of DS among those with ID. **Settings and Design:** This is a community-based study with a survey design. **Subjects and Methods:** A door-to-door survey was conducted by trained, community-based rehabilitation workers under close supervision of multidisciplinary team to identify people with ID. A standardized screening instrument National Institute for Mentally Handicapped-Developmental Screening Schedule was used in the survey. All identified ID cases were evaluated by therapists in IDs for diagnosis of ID on developmental screening test and Vineland social maturity scale. Clinical examination was performed by medical doctors for DS on people identified as ID. Only two parents brought their children for further lab investigations at Ashagram Trust, Barwani. **Statistical Analysis Used:** Descriptive statistics was applied manually to treat the data. **Results:** The frequency of DS population in tribal population closely matches with DS prevalence in the USA. Mothers of DS children in the tribal community are relatively younger. **Conclusion:** Prevalence of DS in tribal population of India may greatly vary with that of the US data, but it is markedly associated with younger maternal age. Further studies are needed for prevalence and identification of potential correlates of this condition.

**Key words:** Down syndrome, genetics, India, intellectual disability, tribal

## Introduction

Down syndrome (DS) is one of the leading genetic causes of intellectual disability (ID) in the world.<sup>[1]</sup> DS alone accounts 15–20% of ID population across the world.<sup>[2]</sup> This condition arises from certain types of disturbance in genetic mechanism that leads to the development of an extra chromosome. DS are classified into three main categories. Trisomy, which is the most common type and accounts for 95% of the total DS population. Translocation and mosaic are less prevalent; these accounts for 3% and 2%, respectively.<sup>[3]</sup> People with DS

are susceptible to various chronic disorders, infections, and disabilities. Among all developmental disabilities such as Autism, Attention Deficit Hyperactive Disorders, and IDs, the IDs are highly prevalent in DS population. According to the studies conducted in the Western countries, 75% DS population suffers from hearing loss, 50–75% with sleep disorders and ear infection, 60% eye disease, 22% psychiatric disorders, and 50% from heart diseases.<sup>[4–6]</sup> The frequency of DS was observed 1 per 1150 in a survey of 94,910 newborns in three metropolitan cities of India: Mumbai, Delhi, and Baroda.<sup>[7]</sup> Genetic disorders including DS are becoming a common cause of mortality in urban newborns.<sup>[8]</sup> Cross-cultural studies have shown consanguineous marriages, high birth rate, and advance maternal age as well of fathers' at conception have a greater risk for DS.<sup>[1,8,9]</sup> Chemical exposure to the parents, radiation

### Address for correspondence:

Dr. Ram Lakhan, Department of Epidemiology,  
Jackson State University, Jackson, MS, USA.  
E-mail: ramlakhan15@gmail.com

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during pregnancy, and socioeconomic factors - parental education and place of living, and risk behavior of fathers' such as smoking are potential attributes of DS aneuploidy.<sup>[10-12]</sup> Because tribal population suffers with higher social, biological, and environmental risks of DS compared to rural and urban population in India, there is a chance this population might show higher prevalence rate of the condition. Estimation of DS prevalence in the overall population is highly needed in India for planning of services and management purpose. This need is higher for tribal and rural population that faces scarcity of genetic counseling, rehabilitation, medical, educational, and welfare services.<sup>[8]</sup> However, there is a need for robust data from Indian population. Therefore, this study was conducted to describe the profile and frequency of DS population of a tribal village and compare it with global data.

### Aim and objectives

The study aimed to understand the profile of DS in tribal population with two objectives: (1) To find the prevalence of DS in tribal population and (2) its comorbidity with ID in tribal population.

### Subjects and Methods

The study was conducted in Chikhalia, a tribal village in Barwani Block of Barwani district in the state of Madhya Pradesh, India. This village has 99.9% tribal population. Approximately, 95% population of this village lived below the poverty line in 2000. Ashagram Trust (AGT), a nongovernment organization, provides services to the people with ID and mental illnesses of this village under a community-based approach. This village is geographically spread in a radius of 4 km and had a population of 2767. The village was selected based on the prior work of the first author who worked extensively in this area in association with the said organization, which is running several community-based programs and has a good rapport with community. The village was surrounded by six tribal villages (Vedpuri, Menimata, Temla, Sindhikhodri, Rasgoan, and Hirachrai) in all four directions and most of the houses of this village were approachable through motorcycle or up to half an hour walk from the place where path ended for the motorcycle. A door-to-door survey was conducted by the community-based rehabilitation workers under the close supervision of therapists in IDs for identification of people with ID. A standardized valid and reliable instrument, "National Institute for Mentally Handicapped-Developmental Screening Schedule" was used in this survey.<sup>[13]</sup> Children identified as ID in screening survey were assessed by two therapists in ID

on standardized tests developmental screening test and Vineland social maturity scale for the diagnosis purpose.<sup>[13]</sup> The comorbid conditions were also assessed by the same ID professionals and also by a psychiatrist for medical conditions such as epilepsy and psychiatric disorders. Clinical/physical examination is considered a first and most sensitive method for the provisional diagnosis of DS.<sup>[4,14]</sup> Children diagnosed as having DS on the basis of clinical examination by an ID professional (first author) and also by a psychiatrist were referred to the medical lab situated in AGT hospital at AGT campus, Barwani. The ID professional was assigned by the AGT to this village for providing and arranging comprehensive services. Two out of four parents were able to bring their children for lab testing. One parent migrated to the Gujarat for seasonal employment and was away from that village for over 6 months. Their blood samples were collected by a licensed lab technician in AGT hospital medical lab and sent to a tertiary laboratory-based hospital, in Baroda, with the help of attending physician in AGT hospital for chromosome evaluation. Children diagnosed as DS are discussed in this paper in reference of the population of that village in that year. The prevalence rate of this population is calculated manually.

### Results

Table 1 provides the profile of children with DS. First three cases (1, 2, and 3) were identified in the survey, while one child (case ID number 4) was identified in the process of community capacity building exercise within the community. The frequency/prevalence of DS ( $4/2767 = 0.00145$ ) equals ( $0.00145 \times 1000$ ), 1.45/1000 in tribal population. Considerably, all mothers of all identified DS children were in their young age (18–24 years); when they had babies with DS.

A separate analysis also indicates that 75% children were on the first birth order. None of the parents of children with DS had consanguineous marriage or history of DS, ID, or any other neurological disorder such as cerebral palsy and epilepsy in preceding generation.

**Table 1: Profile of children with DS**

Case ID	Age (years)	Gender	Birth order	Mother's age at birth	Diagnosis/ method	Level of ID
1	6	Male	First	20	Trisomy/lab confirmed	Moderate
2	8	Male	First	18	Clinical observation	Moderate
3	14	Female	Third	24	Clinical observation	Severe
4	3	Male	First	22	Trisomy/lab confirmed	Mild

ID: Intellectual disability, DS: Down syndrome

## Discussion

Frequency of DS in the tribal population of this village was observed slightly higher than the reported prevalence rates (0.81–1.2/1000 live births) in India.<sup>[15-18]</sup> However, this frequency closely matches with the US prevalence for DS (1.43/1000 person).<sup>[19,20]</sup> Interestingly, against the odd, mothers of these children were relatively younger, while the DS prevalence highly associates with advance maternal and maternal grandparents' age in the population.<sup>[15]</sup> Studies conducted on urban population of Hyderabad and Mumbai city and the state of Punjab found a similar association of younger maternal age (mean age 30 years) with DS.<sup>[15,21-25]</sup> We also observed that 75% children were on the first birth order. None of the parents of these DS children had consanguineous marriage or history of DS, ID, or any other neurological disorder such as cerebral palsy and epilepsy in preceding generation. Then, it is very intriguing to further explore the factors that contribute to the condition. There is very limited knowledge exists about the prevalence and determinates of DS in India for tribal as well nontribal population for comparing the findings of this study. Therefore, it is hard to say that tribal population has a higher prevalence rate of DS. However, it is known that tribal population is highly impoverished and disadvantaged in several ways and suffers proportionately higher burden of nutritional and genetic disorders,<sup>[26,27]</sup> which are potential factors for DS and because of those factors, this population may be having a higher rate of DS in India. Consanguineous marriages, high birth rate, advanced age of parent/s, chemical exposure, and second-hand smoke may be other contributing factors in tribal.<sup>[1,8-12]</sup> However, this study has specific limitations, which need to be carefully considered before generalizing the findings: A small sample size is the main limitation of this observation. In tribal population, people are not well accustomed for keeping the record of their age. Hence, age of the mothers and children is based on the best estimation. This estimation was done by talking with individuals and also their family members and referring the time with local events and factors, such as who was the village *Sarpanch* of that village at that time. This population transit heavily within and surrounding districts, as well seasonally migrate for employment to nearby states: Gujarat and Maharashtra, which may be another confounder associated with this observation.

## Conclusion

Prevalence of DS in tribal population of India may greatly vary with that of the US data, but it is markedly

associated with younger maternal age. Further studies are needed for prevalence and identification of potential correlates of this condition.

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## Conflicts of interest

There are no conflicts of interest.

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