

Situation analysis of Down Syndrome among newborn at a tertiary level Hospital in Bangladesh

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Newborn, caesarian section, Normal vaginal Delivery, Down Syndrome, Karyotyping

ABSTRACT:

Objectives: This study aims to estimate the burden of health services related to Down Syndrome

Methods: This is a cross sectional study designed to assess the situation of Down Syndrome(DS) at tertiary level hospital among inborn attending neonatal outpatient and inpatient department of Dhaka Medical College.

Result: Frequency of Down Syndrome is about 25(0.21%) among 11,372 deliveries in Dhaka Medical College Hospital by caesarian section and normal vaginal delivery.

Conclusion: Down Syndrome is a tolerable burden for the government to create facilities for early holistic intervention which improve their health, education and near normal life.

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Background of the study

As of 2023, the worldwide disease prevalence scenario is shifting from communicable to non-communicable diseases. Down syndrome which occupies more than 70% of congenital neurodevelopmental disorders with or without comorbidity represents an important non-communicable disease in the pediatric age group. It is the most frequent live-born chromosomal aneuploidy in humans¹. Also, it is the most common genetic abnormality of wide-spectrum intellectual disability and physical abnormalities². The overall incidence of Down syndrome worldwide is reported to be 1/600 to 1/800 live births³. The incidences in India is 1 per 850-900 live births. Down Syndrome affects approximately

23,000-29,000 children born in India every year⁴. There are several reports which exhibit the increased incidence of Down syndrome in different regions of the world with respect to ethnicity, maternal age, race, socio-cultural and economic status, environmental factors, national health policy, holistic support system, and awareness. A rise in maternal age, mostly above 35 years, is shown to increase the incidence drastically to 1/400. The incidence rises to as high as 1/12 by the age of 50⁵. Though the definite cause behind the increased risk of aneuploidy is uncertain, most reports suggest a strong relationship of Down syndrome with advanced maternal age⁶. In addition, mothers under the age of 30 years who have had a previous child with Down

syndrome or other aneuploidy conception (trisomy 13, 18 or 21) have a higher risk, as high as 8-fold of the age-related risk of trisomy 21, and a 1.6- to 1.8- fold increased risk for variable trisomies⁶. Effective antenatal screening programs and prenatal diagnostic services have been implemented in many countries in case of high-risk pregnancies concerning women above 35 years of age, for early detection of such anomalies⁷. Some expert reports suggest, there exists 15% greater risk from age-related factors alone⁸. Another report evaluated, there was no increased risk with increased parity and gravidity after adjustment for age. But a recent study suggests a trend towards increasing risk of Down syndrome with increased parity in both younger and older mothers⁹. Recurrence of aneuploidy is a very rare event. Some experts considered two unrelated Kuwaiti families each having 3 siblings with trisomy 21⁹. They proposed that some genetic predisposition and/or gonadal mosaicism could be the underlying cause of recurrent aneuploidy in these parents leading to an increased risk of recurrence of such pregnancies⁹. A recent study stated that more mothers in the younger age group would indicate a possible genetic predisposition in that population¹⁰. Bangladesh does not have any epidemiological study to state how many people are suffering from the Down syndrome and related risk factors. According to the Bangladesh government's ongoing Disability Identification Survey (DIS), so far, 6058 people have been identified with DS in Bangladesh. Experts say this data is highly unreliable. But organizations working in the similar issues assumes at least 200,000 people with DS are crying in silence and is thus of major socio economic concern¹¹. In our country lack of availability of statistical information from the newborn registry lacks the data on the year-wise maternal age of all the mothers, thus, computation of a risk assessment for Down syndrome is not possible. On the other hand, most hospitals in Bangladesh do not have any facility to confirm the diagnosis of Down syndrome using genetic sequencing. Thus, we often rely on clinical diagnosis of Down syndrome. To achieve 30% of SDGs 3.4 by 2030 the government of the People's Republic of Bangladesh needs to take all the necessary steps for Prevention and Control of Non- Communicable Disease. The present study is aimed to detect the incidence of Down syndrome for policy making and implementation to ensure the quality of life among this population.

Rationale for the study

Down syndrome is popularly known as trisomy 21, because it is a genetic disorder caused by a presence of a complete or part of a third copy of chromosome 21. Most of the hospitals in Bangladesh do not have any facility to confirm the diagnosis of Down syndrome using genetic sequencing. Thus, we often rely on clinical diagnosis of Down syndrome. It is true many people are left undetected and many have been detected wrongly. We have found many people with DS who have been identified as persons with autism or cerebral palsy or even as intellectually disabled and others. As Bangladesh belongs to the Lower MIC, major portion of the country lacks rehabilitation efforts leaving DS patients unidentified thus, they often become victims of exclusion, abuse and live a miserable life. On the other hand, our society is not ready to include and recognize DS children. Even reputed institutions, highly educated professionals show utter disdain towards these children. They cannot even imagine that these children can be educated in the same classroom with other pupils. By expanding our awareness to the grassroots level, we need to question, challenge and change this perception. If children with DS are detected early, they can be taught language and mobility within three years with the help of doctors, speech therapists and occupational therapists. If this can be done within three or four years alongside ensuring the medical care for their characteristic health complications, these children can fully participate in the inclusive education program and lead a normal life. Actually, we need initiative from the government as well as different steps from private organizations regarding policy making and its implementation. Firstly, we have to determine the demographic information of DS in Bangladesh. By taking into account the public health importance of DS, this study aims to detect the incidence and seek for correlation between the related factors in context and the socio-demographic status persistent in Bangladesh. This study will provide a basis for further epidemiological surveys of Down Syndrome in Bangladesh. Also helps to prepare Down syndrome medical pathway for proper management and prepare budget according to the burden of this non-communicable disease by government.

Methods

This is a cross sectional study designed to assess the

situation of Down Syndrome (DS) at selected hospital. The data collection for the study is conducted over a twelve-month period from January 2023 to December 2024. To ensure effective identification of Down Syndrome cases among newborns key personnel (Doctors, Nurses, staffs) was chosen and trained them for proper identification and clinical suspicion of Down Syndrome cases among newborns delivered 4320 by normal vaginal delivery and 7052 by caesarian section at Dhaka Medical College Hospital.

Results

During the study period total babies born about 11,372. Among them 4320 by normal vaginal deliveries in normal delivery facilities of DMCH and in caesarian OT during preparation and 7052 by caesarian section (Table 1).

Table- 1: Number of newborn babies in DMCH during study period (July 2023 to June 2024)

Newborn babies	in labor room	in CS OT	Total
Number of delivery by NVD	4290	30	4320
Number of delivery by Caesarean section		7052	7052
Total newborn			11,372

Table-2: Frequency distribution of Down Syndrome in DMCH (n=11,372)

	Number	Frequency(%)
Newborn without Down Syndrome	11,347	99.79
Cases with Down Syndrome	25	0.21

Among the 11,372 newborns 25 (0.21%) were diagnosed cases of Down Syndrome.

Table-3: Distribution of chromosomal abnormalities according to gender (N=25).

Gender	Free Trisomy	Robertsonian Translocation	Mosaic	Total
Male	14	2	2	18
Female	5	2	0	7
Total	19	4	2	25

Discussion

Based on the research conducted in Dhaka medical college, it shows that total number of newborn delivered in Dhaka Medical College Hospital during the study period is 11,372. Among them clinically suspected Down syndrome cases were thirty, (30) who were examined initially during postnatal caring immediate after birth by specially trained nurses. Trained medical officer working in neonatal outpatient and inpatient department re-examined the babies and selected for further evaluation of Down syndrome. Three babies died due to severe congenital anomalies immediately after birth. Blood samples were collected by trained personnel from 27 babies and send for Karyotyping. Twenty-five (25) cases were confirmed Down syndrome by karyotyping and two (02) report revealed normal. Majority of patients were males (n=18; 72 %). Nineteen (19) cases revealed free trisomy, fourteen (14) cases out of them were male (47XY, +21) and five (05) cases were female (47XX, +21). Out of four (04) cases with Robertsonian translocation, two (02) cases were male and two (02) cases were female. Two (02) cases with mosaicism were male. Among the 11,372 newborns 25 (0.21%) were diagnosed cases of Down Syndrome. It may be due to presence of fetomaternal service in DMCH which leads to increase number of deliveries of high risk mother.

Limitation

This is a single centered and short duration study.

Conclusion

Down Syndrome in newborn is about 25(0.21%) among 11,372 deliveries in Dhaka Medical College Hospital by caesarian section and normal vaginal delivery which resembles the frequency of DS is quite high in related to other studies done in different countries. DS is a tolerable burden for the government to create facilities for early holistic intervention which improve their health, education and near normal life, can also reduce the familial burden. Cytogenetic analysis is the most important diagnostic tool for DS. Cytogenetic data can provide a basis for the genetic counselling of families with DS children and their plan.

Recommendation

To detect the incidence of Down syndrome in Bangladesh and to ensure early detection and management of DS a multicenter long duration study can be done.

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