

**INCIDENCE OF DOWN'S SYNDROME AND ASSOCIATED RISK FACTORS AT MCH,
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ABSTRACT

Introduction: Down syndrome (DS), a congenital condition caused by trisomy of human chromosome 21 is the most frequent chromosomal abnormality in live births associated with mental retardation and congenital heart defect. Worldwide screening strategies, introduced, has helped infants with Down syndrome, have high survival, and better care especially of cardiovascular malformation. (Epstein et al., 2001). **Methodology:** It was a retrospective observational study using a Cross-sectional design, carried out from January- June 2013. The cross sectional study was conducted at the Maternal and Child Hospital (MCH) in Buraydah Qassim, K.S.A. Data was abstracted from medical records of neonatal babies born, during the period of January to December 2012, using the consecutive sampling method. For each down syndrome case, two normal control cases were randomly selected, within the ascertained period January to December 2012. **Results:** Total number of babies (mothers), enrolled in the study were, 33. The number neonates having Down Syndrome was 11, while number of normal controls was 22. Mean ages of mothers of down syndrome was of 33.6 years versus 29.3 years for mothers of normal children ($P = .129$) and 63% ($n = 7$) ≥ 36 years). Analysis of the demographic factor, previous exposure to radiation, showed a mean \pm SD 1.9091 \pm .30151, with a significant (p value = 003*). Analysis of mothers, who had diabetes showed a mean \pm SD 1.6364 \pm .50452, with a (p value=.001*), which was significant. **Conclusion:** The Incidence of Down syndrome (DS) in Maternal & Child Hospital (MCH) in Buraidah, Al Qassim, during the period of January 2012 – December 2012, was 11 per 10,000 deliveries. The most common **maternal** demographic factors, found to be associated with Down Syndrome(DS) were increased maternal age and increased occurrences of Down Syndrome, Association of Diabetic mother's with Down Syndrome, and Association of maternal exposure to radiation with Down Syndrome. **Neonatal** demographic factors, did not show any significant association of low birth weight, sex of child, or other demographic factors studied. The only comparative association, was of a lower Total Apgar score.

KEYWORDS: Down Syndrome (DS), Maternal & Child Hospital (MCH).**INTRODUCTION**

Down syndrome (DS), a congenital condition caused by trisomy of human chromosome 21 is the most frequent chromosomal abnormality in live births associated with mental retardation and congenital heart defect (Epstein, 2001). Down syndrome screening has been an integral part of routine prenatal care in the recent decades. In Saudi Arabia the diagnosis of Down syndrome is made by chromosomal analysis which can be initiated prenatally due to given risk factors for pregnancy of certain women, or postnatal due to the characteristic appearance of the newborn child. Currently, there is no established national antenatal screening strategy for Down syndrome in the Kingdom of Saudi Arabia.^[9]

The characteristic features and symptoms of Down syndrome(DS)

Severity of DS ranges from mild to severe, most individuals with DS, have widely recognizable physical characteristics. These include: a flattened face and nose, a short neck, a small mouth sometimes with a large, protruding tongue, small ears, upward slanting eyes that may have small skin folds at the inner corner (epicanthal fold). White spots (also known as Brushfield spots) may be present on the colored part of the eye (iris); the hands are short and broad with short fingers, and with a single crease in the palm; poor muscle tone and loose ligaments are also common; and development and growth is usually delayed and often average height and developmental milestones are not reached.

Risk factors

1. Advancing maternal age. 2 Having had one child with Down syndrome. 3. Being carriers of the genetic translocation for Down syndrome. Complications, include Heart defects, Leukemia, Infectious diseases. Dementia, Sleep apnea, Obesity & Other problems, as gastrointestinal blockage, thyroid problems, early menopause, seizures, hearing loss, premature aging, skeletal problems and poor vision.

Diagnosis of Down syndrome can be done, before birth using one of several diagnostic tests such as;

Amniocentesis is performed between 16 and 20 weeks of pregnancy. The sample is analyzed for chromosome anomalies.

Chorionic villus sampling (CVS) is done between 11 and 12 weeks of pregnancy. The chromosomes in CVS are analyzed for deviations.^[9&10]

For percutaneous umbilical blood sampling (PUB): The blood sample is examined for chromosome abnormalities. It is usually performed after week 18. **Karyotype analysis** usually requires seven to 10 days. A recently developed assay that uses fluorescent in situ hybridization (FISH) can allow rapid diagnosis of trisomy 21 after amniocentesis.^[9] Ultrasound Screening during pregnancy is also helpful for diagnosis.

MATERNAL SERUM SCREENING: Maternal serum screening (multiple- marker screening) can allow the detection of trisomy 21 pregnancies in women in younger age group^(3&4). Alpha-fetoprotein (AFP), unconjugated estriol and human chorionic gonadotropin (HCG) are the serum markers most widely used to screen for down syndrome.^[5] this combination is known as the "triple test" or "triple screen. With trisomy 21, second-trimester maternal serum levels of AFP and unconjugated estriol are about 25 percent lower than normal levels and maternal serum HCG is approximately two times higher than the normal HCG level.^[4] The triple test is usually performed at 15 to 18 weeks of gestation. A standard risk cutoff is used to determine when the test is considered "positive, cutoff of 1/270, is equal to the second-trimester risk of trisomy 21 in a 35-year- old woman.^[5] the triple test can detect 60 percent of trisomy 21 pregnancies; it has a false-positive rate of 5 percent.^(3&6)

Treatment includes, Rehabilitation and Corrective surgery for heart defects, gastrointestinal irregularities, and other health issues, becomes essential for some individuals. Regular health checkups should be scheduled to screen for other conditions such as visual impairments, ear infections, hearing loss, hypothyroidism, obesity, and other medical conditions.

OBJECTIVES

General objective; To assess the factors that associated with Down syndrome at MCH in Buraydah.

Specific objectives

To collect the medical records Babies (Mothers) in obstetric department at MCH from Jan to Dec 2012.

To determine no. of babies with DS and without DS during this period.

To assess the Baby's Birth History of normal child and down syndrome child

To assess maternal demographic, medical & obstetric history.

To assess prenatal & postnatal history of the baby.

METHODOLOGY**Study Design**

This was an observational study using the Cross-sectional study design method carried out from January to June 2013. The cross sectional survey was conducted at the Maternal and Child Hospital (MCH) in Buraydah Qassim Reigon K.S.A. Data was abstracted from medical records of neonates born during the period of January 2012 to December 2012.

Study Sample

The consecutive sampling method was utilized to abstract information from the medical records of neonates born during the period of January to December 2012.

Sample Size: The availability of complete data of, newborns in (2012) from medical records at the MCH hospital, in Buraydah, Al- Qassim, was the criteria to include in this study. For each DS case two normal control cases were randomly selected, within the ascertained period January to December 2012.

Inclusion criteria: Infants born at MCH during Jan-Dec 2012, with complete data relevant to this study.

Exclusion: Infants born after Dec 2012, who did not have complete data for this study.

Data Collection; Informed consent were obtained from the relevant authorities, for the conduct of this study and in particular from MCH for the medical record abstraction and data collection. The information obtained for this research from MCH, was used without any identifiers of individual patients to maintain their confidentiality. All information obtained from the patient medical records was used for research purposes only. A structured questionnaire was prepared and used as a Performa to collect information on Mother's demography (age, number of pregnant, birth order, family history, consanguinity), Mother's obstetric history, baby's birth history (type of delivery, Apgar's score, diagnosis of down syndrome, birth weight, length, head shape).

Statistical Analysis: The statistical analysis of the data was carried out by using the Statistical Package for

Social Science (SPSS version no.20) to determine the Incidence of down syndrome. Descriptive statistics was used to examine the distribution of each study variable individually with mean, median and standard deviation.

RESULTS

The present study was done at Maternal and Child Hospital (MCH) in Buraydah Qassim reigon, K.S.A, in June 2013. Thirty three medical records of neonates born during the period of January to December 2012 were included in this study. Many characteristics of mother and neonates were studied, to find the associations of this characteristics with down syndrome.

Table1: Associations of Mother characteristics with Down Syndrome cases.

Mother characteristic	DS Case Mean± SD	Normal Control mean±SD	p.value
Mean.Age	36.4545 ±8.02949	29.3182 ±5.53247	.129
Mean.WT	73.0909 ± 11.62931	69.6364 ±8.79824	.122
Mean.HT	158.5455±4.82418	159.0909±5.53697	.603
Mean.Birth. Order	4.6364±2.73030	3.2273±1.87545	.152
Mean.Previous Medical History	2.8182±1.53741	3.5000±2.28348	.097
Mean.RAD. Exp.	1.9091±.30151	2.0000±.00000	.003*
Mean.Diabetic Mothers	1.6364±.50452	1.9091±.29424	.001*
m.MED.	2.0000±.44721	2.2273±.75162	.272

*(p<0.005)

The number of noenates having down syndrome was 11, and their mother,s mean ages, was 33.6 years versus 29.3 years for mothers of normal children ($P = .129$) and 63% ($n = 7$) ≥ 36 years). Their mean weights was 73.1 kg, compared to mothers of normal children whose mean weights was 69.6 kg, which did not have a significant p value. Mothers of DS children had a mean parity of ≥ 4.6 versus 3.2 ($P \geq .152$) which was not significant. Statistical analysis of mother's previous history (previous c/s 4xGA no complication, G3P2 SVD, hypertension, infection, DM family history, abortion, asthma) their mean \pm SD was 2.8182 ± 1.53741 . Analysis of the demographic factor, previous exposure to radiation had mean \pm SD $1.9091 \pm .30151$, with a significant(p value = 003^*). Analysis of mothers that had diabetes showed a mean \pm SD $1.6364 \pm .50452$, with a (p value= $.001^*$), which was significant. Other maternal demographic factors related to Down syndrome were not significant.

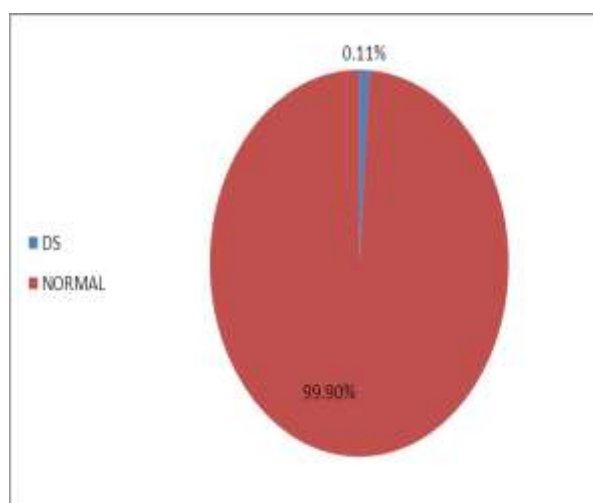


Figure 1: Incidence of DS at MCH in Buraydah, Qassim, K.S.A. 11/ 10,000 population.

Table 2: Associations of Neonatal characteristics with Down Syndrome.

Neonatal characteristics	DS Case Mean± SD	Normal Control mean±SD	p.value
n.SEX	1.5455±.52223	1.4545±.50965	1.000
n.TOD	1.3636±.50452	1.3182±.47673	.634
n. T. APGAR	24.0000±2.19089	27.9545±1.46311	.946
n.BirthWT.	2.9273±.62465	2.9268±.43415	.438
n.Length.	48.7273±4.17351	48.6364±4.35940	.913
n.Head Circum	33.7273±1.53889	34.2955±1.27857	.927

The Incidence of DS was 11 per 10,000 live births annually. Compared with the reference control group, the 11 children with trisomy 21 had a mean birth weight of 2.927 g versus 2.9268, ($P = 0.438$).

The mean for sex with DS was 1.5 versus 1.4 in normal children ($p \geq 1.000$), which did not show significant co-relation to sex. The mean of Total Apgar score (At birth

+ After1 minute + After 5 minute = Max. 30) in DS was 24, compared to mean T. Apgar score of 28 in normal children ($p \geq .946$) which was combatively less, but not significant. Other parameters evaluated were, c. length, c.Head circumference, c. Type of delivery (TOD) between DS & Normal children, which did not show any significant co-relation. ($p \geq 0.005$).

DISCUSSION

The epidemiology of Down syndrome (DS) was studied in the Maternal & Child Hospital (MCH) Buraidah in Qassim From Jan - June 2013, which studied 33 medical records of neonates born during the period of January to December 2012, extracted from Medical records section MCH. For each DS case two normal control cases were randomly selected, within the ascertained period January to December 2012. More than 20 factors were studied and compared to those from control infants. The Incidence of DS was 0.11%; 11 DS cases out of 10,000 deliveries during the mentioned time period.

In our study interesting maternal demographic factors found co-related with DS were;

1. The increasing mother's age and occurrence of down syndrome; as the mean ages of mother's of DS was 33.6 years versus 29.3 years for mothers of normal children ($P = .129$), and 63% ($n = 7$) of these mothers were ≥ 36 years. Though this co-relation was not having a significant p value, but it does indicate a positive relationship. Other studies from Saudi Arabia & outside also show the same co-existence.^[9&10]
2. Mothers of DS children had a mean parity of ≥ 4.6 versus 3.2 ($P \geq .152$) which was not significant, in our study due to a small sample size, but this positive co-relation has been proved by other studies.^[7]
3. Analysis of mothers that had diabetes showed a mean \pm SD 1.6364 \pm .50452, with a (p value= $.001^*$), which was significant. Other studies have proved, a high incidence of Down's syndrome in infants of diabetic mothers; The incidence of Down's syndrome was studied in 1870 infants of diabetic mothers out of 22 300 neonates born between January 1987 and April 1994 by Narchi et al. All pregnancies were screened for diabetes and all cases of Down's syndrome were confirmed by chromosome analysis. Down's syndrome (all trisomy 21) was diagnosed in 35 infants: seven were born to mothers with gestational diabetes and 28 to non-diabetic mothers. The incidence of Down's syndrome was higher in infants of diabetic mothers (3.75 per 1000 v 1.36 per 1000) ($p = 0.02$) with a relative risk of 2.75.^[11]
4. Analysis of the demographic factor, mother's previous exposure to radiation had mean \pm SD 1.9091 \pm .30151, with a significant (p value = 003^*). As such no other study has reported a significant relationship between, mother's exposure to radiation and occurrence of down syndrome, so this might be suggesting; a high exposure to radiation in the child bearing age group, being practiced as routine investigation protocol in this region. As our study group was small, this co-relation has scope to be reviewed further. Other maternal demographic factors related to Down syndrome were not significant.

The most interesting neonatal factors, associated with DS, found in our study, were;

1. The mean birth weight of 2.927 g in DS versus 2.9268 in Normal children, ($P = 0.438$). Our study did not report a positive relation between low birth weight & DS, whereas other studies have shown a significant relation of L.B.W & DS ($P < .001$)^[7]
2. The mean for sex with DS was 1.5 versus 1.4 in normal children ($p \geq 1.000$), which did not show significant co-relation to sex, and the same has been reported by other studies.^[7]
3. The mean of Total Apgar score (At birth + After 1 minute + After 5 minute = Max. 30) in DS was 24, compared to mean T. Apgar score of 28 in normal children ($p \geq .946$) which was comparatively less, but not significant.
4. Other parameters evaluated were, c. length, c. Head circumference, c. Type of delivery (TOD) between DS & Normal children, which did not show any significant co-relation. ($p \geq 0.005$), though some studies have given a significant association for c. Head circumference & DS. At birth, the DS infants measured and weighted less and their head circumference was lower than in control infants.^[8]

For all other factors studied no statistically significant difference with respect to controls could be demonstrated.

CONCLUSION

The Incidence of Down syndrome (DS) in Maternal & Child Hospital (MCH) in Buraidah, Al Qassim, during the period of January 2012 – December 2012, was 11 per 10,000 deliveries. The most common **maternal** demographic factors were; Increased maternal age, High parity of mother, Diabetic mother's and maternal exposure to radiation. **Neonatal** demographic factors, did not show any significant association of low birth weight, sex of child, or other demographic factors studied. The only comparative association, was of a lower Total Apgar score. There are need for more studies, in and around Buraydah Al-Qassim, Saudi Arabia as, our sample size was small and limited time period of the study. Routine screening of all expecting mothers above the age of 35 years by the Triple test & karyotyping techniques, for early detection, and public education programs, & a National Rehabilitation & Vocational Safety program for DS cases is recommended.

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