

Risk factors and parent's knowledge related to Down syndrome: A retrospective study.

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Abstract

Down syndrome or Trisomy 21 is the most common chromosomal abnormality. It is caused by chromosomal malfunction during the meiosis process. Some parents are more likely to have a child with Down syndrome. Risk factors include advancing maternal age and carrying the Down syndrome genetic translocation. Treatments are based on each individual's physical and intellectual needs as well as his or her personal strengths and limitations. Medical treatment can be used to treat illnesses including seizure disorders, hypothyroidism, and juvenile leukemia. Several studies investigated possible risk factors for Down syndrome and sex chromosomal aneuploidy. In the current study a simple random sample will be utilized to choose participants based on inclusion criteria for this study, and all people who agree to participate will be included in the study. A questionnaire will be used by the research team to collect data. Data monitoring, safety considerations, handling complications, samples disposal, and hazardous materials have been examined.

Keywords: Down syndrome, Trisomy 21, Chromosomal malfunction, Questionnaire, Data monitoring.

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Introduction

Down Syndrome (DS) or Trisomy 21 is the most common chromosomal abnormality. The non-separation or Non-Disjunction (NDJ) of Chromosome 21 (Ch21) during gametogenesis results in disomic gametes having two copies of a given chromosome, which, when fertilized by a haploid gamete of the opposite sex, result in the production and implantation of a trisomic baby. However, of all autosomal trisomies, it is also the most compatible with survival. Mongolians or Mongols are the terms used to describe people who have Down syndrome. Down syndrome affects children of all ethnic backgrounds. This is caused by chromosomal malfunction during the meiosis process, and it is more frequent in older parents Chicoine et al. [1]. According to prenatal statistics, 1–2.2 of every 1,000 live babies are affected Enea et al. [2].

Some parents are more likely to have a child with Down syndrome. The following are some of the risk factors: advancing maternal age, being carriers of the genetic translocation for Down syndrome, having had one child with Down syndrome [3]. Because older eggs have a higher risk of incorrect chromosomal division, a woman's odds of having a kid with Down syndrome grow with age. After 35 years of age, a woman's chances of having a child with Down syndrome rise. However, because younger women have considerably more infants, the majority of Down syndrome children are born to women under the age of 35 [4].

Both males and females can carry the Down syndrome genetic translocation to their children. Parents who already have one kid with Down syndrome or who have a translocation are more likely to have another child with Down syndrome. A genetic counselor can assist parents in determining the likelihood of having another child with Down syndrome [5]. In certain

countries, the number of infants born with Down syndrome has more than doubled as maternal age has grown. Even in nations where abortion is legal, it is becoming increasingly prevalent [6] as a result of the widespread use of prenatal diagnostic methods [7]. There are a number of Down Syndrome Screening (DSS) tests available, with rates of prediction obtained from a single test or a combination of tests, giving you a number of options.

The non-invasive prenatal diagnostic is the most recent technological advancement is that analyses free circulating fetal DNA in the mother's blood. It has a strong predictive value, making it an excellent candidate for secondary screening following a positive classical DSS to avoid invasive treatments [8,9]. Although the genetic influence of advanced father age on Down syndrome is widely documented, the genetic impact of advanced mother age is less well understood. Older dads, like older mothers, may have a role in the higher risk of Down syndrome among children born to older couples. However, more research into the impact of maternal age on the development of Down syndrome is needed [10,11]. Four life stages of Down syndrome were identified: prenatal, childhood, adulthood, and senescence. Pneumonia and other viral infections were the commonest cause of death across the entire lifespan of individuals with Down syndrome [12,13]

Although there is no treatment for Down syndrome, there are a number of treatments and therapies that may be used to address a child's specific medical problems and developmental characteristics. Treatments are based on each individual's physical and intellectual needs as well as his or her personal strengths and limitations [14]. Medical treatment can be used to treat illnesses including seizure disorders, hypothyroidism, and juvenile leukemia, depending on the child's underlying medical difficulties. Upper neck anomalies and gastrointestinal troubles

may necessitate surgery [15]. As well, a wide range of treatments are available to meet a child's particular physical, behavioral, and communicative requirements, and they may have a substantial positive influence on the learning and development of children. Moreover, educational therapy used to treat the main symptoms of Down syndrome is given through state and local school systems [16].

Due to the lack of information and studies about Down syndrome, this study aims to: to assess the level of knowledge and awareness regarding to Down syndrome, risk factors of Down syndrome, to know if participants were conducted Nuchal Translucency (NT) test during pregnancy and to compare if there any statistical association between down syndrome and some demographic characteristics and health history of patients.

Literature review/background

A cross-sectional investigation was done between March 2006 till February 2008. Shalaby et al. shed some light on other potential Down syndrome risk factors. They stated that, forty-eight clinically suspected cases with Down syndrome ranging 6 months to 9 years were submitted for chromosomal investigation (karyotyping). According to the data, 45 of the 48 Down syndrome cases had free trisomy 21, two had mosaic trisomy 21, and one had translocation. By using logistic regression on a case-control study of Down syndrome children, the odds ratio of uncle-niece marriages, or second cousin marriages, or parents living in rural areas, or parents exposed to drugs and therapies, or parents' education level, or father's behavioral patterns (cigarettes/coffee use), or mothers not having undergone ante-natal checking as part of post-natal care, or mothers with a Down syndrome child was demonstrated. Moreover, they reported that consanguineous marriage, parental residence location (rural/urban), parental chemical exposure, parental educational status, father's behaviors, prenatal scans, and mother's reproductive success are probable risk factors for Down syndrome.

In addition, between 2003 and 2009, conducted a case study of around 400 children with or without Down syndrome, as well as their parents, predominantly in and around Kolkata, India [17]. The study's purpose was to get a better knowledge of the epidemiology of Down syndrome and the intricate interplay between genetic and environmental hazard factors in the oocyte. Microsatellite markers were used to genotype family members in order to pinpoint the source of nondisjunction faults and non-disjoined chromosome 21 recombination designs.

In Mysore, South India, Ramachandra et al. investigated possible risk factors for Down syndrome and sex chromosomal aneuploidy. Over the course of five years, 150 clinically confirmed DS and 25 SA cases from Mysore's top hospitals were sent to their laboratory for chromosomal analysis. These patients' chromosomal preparations were obtained after getting written consent. Well-spread G-banded metaphase plates were examined using the automated LEICA KARYO algorithm. 200 and 100 randomly chosen households from various religions

were used as controls in the DS and SA cases, respectively. In the statistical study, logistic regression was performed. A total of 122 of the 150 DS patients had free trisomy 21, two had mosaic trisomy 21, and one had translocation. According to the study, consanguinity, parental home area (rural/urban), parental chemical exposure, parental education level, father's behaviors, prenatal scanning, and mother's reproductive performance are all potential risk factors for chromosomal aneuploidy [18].

Jan et al. conducted a cross-sectional analytical questionnaire-based study among select Jeddah residents. A total of 360 persons completed the surveys [19]. The responses varied in age from 16 to 70, with 45.2% females and 43.7% men. The average age was 36 years. 67% had attended college, 5% had earned a postgraduate degree, 23.5% had graduated from high school, and 3.1 percent had just finished elementary school. The majority of those who took part had a high monthly salary 38%. They revealed that the persons who were interviewed had a poor grasp of Down syndrome. Furthermore, because education has a significant influence on awareness of Down syndrome, it is necessary to educate the public and patients with the necessary information to help them better comprehend the illness.

Buyukavci et al. conducted a study to analyze the viewpoints of moms of Down syndrome children at the time of diagnosis, as well as the communication attitudes of healthcare practitioners. The research included mothers of children with Down syndrome. The mothers' experiences at the time of diagnosis, as well as the perspectives of healthcare professionals, were assessed using a semi structured interview. The study enlisted the participation of 43 moms. Eight infants were identified with Down syndrome before birth, while 35 children were diagnosed after birth. They reported that mothers of children with Down syndrome need some more information about their children's condition from healthcare practitioners. They also expect this information to be delivered in a warm and caring manner. Healthcare practitioners, particularly pediatricians, must allot adequate time to discuss with families about Down syndrome and its complications.

Deakin et al. conducted a research from the perspective of their mothers to investigate the societal stigma that young people with Down syndrome and their families suffer on a daily basis. Using an interpretative phenomenological technique, the mothers' perceptions of their child's increasing understanding of their disability and associated societal stigma were studied (IPA). According to the data, all mothers were aware of Down syndrome's stigmatized status and went to considerable pains to prevent their children from knowing about it. Down syndrome has been brought up by some moms with their children. Other moms watched their children for signs that they were ready to talk about it. On the surface, all of the mothers believed their children had a limited awareness of Down syndrome and the stigma associated with it, but a more in-depth conversation revealed a more sophisticated understanding. Mothers expressed uncertainty and concern about when and how to talk to their children about Down syndrome.

Stensson et al. analyzed quantitative data from an online questionnaire (Chi-square test). The goal of this online survey was to find out how parents felt about their children's overall health, oral health, and dental health care among Swedish Down Syndrome children (DS). The study included the parents of 101 children with Down syndrome. 70% of parents reported their child's overall health was good or excellent, and 74% said their child's dental health was good or excellent. The parents wished for multidisciplinary collaboration. According to the data, most parents rated their child's overall and dental health as good or very good. Dental procedures have also been highlighted as being problematic for youngsters with poor oral health. Parents sought dental treatment tailored to their child's individual need. They wanted dental workers to be familiar with youngsters who need extra care. Finally, they requested cross-disciplinary collaboration.

Methods

Data collection and analysis

Study population and study area: This research is available to all parents who have a child with Down syndrome.

Sampling techniques and sample size: A simple random sample will be utilized to choose participants based on inclusion criteria for this study, and all people who agree to participate will be included in the study.

Inclusion criteria: All parents who have child with Down syndrome are eligible to participate in this study.

Sample size: Recent study conducted in 2020 by Abdullah Shalabi and his colleagues indicated that 61.4% of females found that the awareness of Down syndrome was moderate, so the sample size was calculated accordingly.

Sample size will be calculated by using this equation:

$$n = z^2 pq / m^2$$

Where:

$z = z$ value (1.96 for 95% confidence level)

$p =$ assumed proportion = 61.4

$q = 1 - p$ (complementary) = 38.6

$m =$ margin of error = 0.05

So the sample size will be $(1.96)^2 * 0.614 * 0.386 / (0.05)^2$ equal 364.

Data collection tool: A questionnaire will be used by the research team to collect data. The questionnaire will be divided into three sections: part one will contain participant demographics and health history, part two will include risk factors connected to Down syndrome, and part three will include variables to assess participants' understanding of Down syndrome.

Management plan: Data monitoring, safety considerations, handling complications, samples disposal, hazardous materials, etc.

Ethical considerations: This study proposal should be approved by the Saudi Arabian ethical committee, and all ethical issues should be considered throughout the study process. All participants should fill out an informed consent form prior to participating, and they should be informed about the study's objectives, as well as their rights to withdraw from the study if necessary. They should also be informed about the confidentiality of the data collected.

Work activity plan (Gantt chart): Attached table showing the detail of the activities by time.

Results

They reported that consanguineous marriage, parental residence location, parental chemical exposure, parental educational status, father's behaviors, prenatal scans, and mother's reproductive success are probable risk factors for Down syndrome (Tables 1 to 3).

Part one: Demographic data
Age in years
Educational level
Primary
Intermediate
Secondary
Diploma
BSc
Post graduate
Marital Status
Married
Divorced

Widow
Height in CM
Weight in Kgms
BMI
Number of children

Table 1. Part one: demographic data.

Part two: Risk factors
Married
Divorced
Widow
Height in CM
Weight in Kgms
BMI
Number of children
1. Age since married in years
2. Age of parent when birth of Down syndrome child
Father Mother
3. Do you have previous abortions?
Yes
No
4. Consanguineous marriage
Yes
No
5. Genetic disease in mother family
Yes
No
6. Genetic disease in father family
Yes
No
7. Received antenatal care
Yes
No
8. Exposure of parents to drugs
Father on medications
Mother on medications
Both on medications
9. Smoking of father
Yes

No
10. Smoking of mother
Yes
No

Table 2. Part Two: Risk factors.

Part three; Knowledge and awareness related to Down Syndrome
1. Do you hear about Nuchal Translucency (NT) test during pregnancy
Yes
No
2. Do you think that the Down syndrome has risks
Yes
No
3. Do you have knowledge about the complication of Down syndrome
Yes
No
4. Do you think Down syndrome is a genetic disorder
Yes
No
5. Do you think Down syndrome leads to severe complication can cause death
Yes
No

Table 3. Part three; knowledge and awareness related to Down syndrome.

Discussion

Down syndrome occurs when a child is born with an extra copy of chromosome [20]. Physical and intellectual difficulties can occur in people with Down syndrome. Down syndrome cannot be treated. Treatment programs can assist persons with the illness improve their abilities and live happy, productive lives. As a woman gets older, her chances of having a kid with Down syndrome rise. Previous studies investigated the potential Down syndrome risk factors.

Conclusion

Our study aimed to gather more information and analyze the amount of knowledge and awareness about Down syndrome, risk factors for Down syndrome, and whether or not individuals had a NT test during pregnancy. It also seeks to determine whether there is a statistical link between Down syndrome and certain demographic features and health history of patients. The study team gathered data using a questionnaire. The questionnaire divided into 3 sections: The first one covered participant demographics and family medical

history, the second included risk factors associated with Down syndrome, and the third included parameters to measure participants' comprehension of Down syndrome.

Conflict of Interest

The authors declare that they have no conflict of interest.

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