

ORIGINAL ARTICLE

Assessment of Knowledge and Awareness of Down syndrome in Ha'il City Community, KSA

AMANY MOHAMMED KHALIFA¹, SHAIMA SAAD ALJOHANI², FAUZIAH ABDULLAH ALSHAMMARI³

^{1,2,3}Faculty of Medicine, University of Ha'il, Ha'il, KSA.

Correspondence to: Amany Mohammed Khalifa

ABSTRACT

Background: Down syndrome is a condition in which a person has an extra chromosome. Chromosomes are small "packages" of genes in the body. They determine how a baby's body forms and functions as it grows during pregnancy and after birth. Typically, a baby is born with 46 chromosomes. Babies with Down syndrome have an extra copy of one of these chromosomes, chromosome 21. A medical term for having an extra copy of a chromosome is 'trisomy.' Down syndrome is also referred to as Trisomy 21. This extra copy changes how the baby's body and brain develop, which can cause both mental and physical challenges for the baby.

The aim: the present study builded the interest to assess the knowledge and awareness of definition, causes, symptoms, diagnosis, complication, and treatment of Down Syndrome (DS) in Hail City and tried to improve the false thoughts about this disease.

Material: A cross-sectional survey was distributed throughout in Hail City between October 2021 to April 2022. An online questionnaire was designed. It included informed consent and 10 questions about the sociodemographic data and questions regarding signs, symptoms, complications and management about DS. It was distributed via various social media apps.

Methods: SPSS for Windows v22.0 IBM Inc..SPSS for windows Rel 15.0 2006 Chicago Inc.

Results: The result of the present study revealed that the overall level of awareness about Down syndrome was moderate (65.7 %). More Health education for public, online seminars and more research are needed to raise the public awareness and spotlight to patients suffering from this syndrome.

Keywords: Down Syndrome, genetic disease, awareness, Ha'il, KSA,

INTRODUCTION

Down syndrome (DS) is a birth defect with huge medical and social costs, caused by trisomy of whole or part of chromosome 21. It is the most prevalent genetic disease worldwide and the common genetic cause of intellectual disabilities appearing in about one in 400-1500 newborns. Although the syndrome had been described thousands of years before, it was named after John Langdon Down who described its clinical description in 1866. Scientists have identified candidate genes that are involved in the formation of specific DS features. These advances in turn may help to develop targeted therapy for persons with trisomy 21. Screening for DS is an important part of routine prenatal care. Until recently, noninvasive screening for aneuploidy depends on the measurement of maternal serum analytics and ultrasonography. More recent progress has resulted in the development of noninvasive prenatal screening (NIPS) test using cell-free fetal DNA sequences isolated from a maternal blood sample [1].

Individuals with Down syndrome (DS) commonly possess unique neurocognitive and neurobehavioral profiles that emerge within specific developmental periods. These profiles are distinct relative to others with similar intellectual disability (ID) and reflect underlying neuroanatomic findings, providing support for a distinctive phenotypic profile. This review updates what is known about the cognitive and behavioral phenotypes associated with DS across the lifespan. In early childhood, mild deviations from neurotypically developing trajectories emerge. By school-age, delays become pronounced. Nonverbal skills remain on trajectory for mental age, whereas verbal deficits emerge and persist. Nonverbal learning and memory are strengths relative to verbal skills.

Expressive language is delayed relative to comprehension. Aspects of language skills continue to develop throughout adolescence, although language skills remain compromised in adulthood. Deficits in attention/executive functions are present in childhood and become more pronounced with age. Characteristic features associated with DS (cheerful, social nature) are personality assets. Children are at a lower risk for psychopathology compared to other children with ID; families report lower levels of stress and a more positive outlook. In youth, externalizing behaviors may be problematic, whereas a shift toward internalizing behaviors emerges with maturity. Changes in emotional/behavioral functioning in adulthood are typically associated with neurodegeneration and individuals with DS are higher risk for dementia of the Alzheimer's type. Individuals with DS possess many unique strengths and weaknesses that should be appreciated as they develop across the lifespan. Awareness of this profile by professionals and caregivers can promote early detection and support cognitive and behavioral development [2].

In a previous study the prevalence rate of Down syndrome in southern Thailand in 2009 was significantly increased with maternal age. About 35% of DS cases were detected prenatally and later terminated. Hence, examining only registry live births will result in an inaccurate prevalence rate of DS [3]. The symptoms were The physical abnormalities that together give rise to the distinctive facial appearance associated with this condition include upslanting palpebral fissures with inner epicanthic folds, flatness of the bridge of the nose, midfacial hypoplasia, and a tendency to protrude the tongue, especially at young. Many other functionally inconsequential minor abnormalities of the ears, hands, and feet might also be present with short stature.

Approximately 40% of affected individuals were born with congenital heart disease, with endocardial cushion and related septal defects frequently being present. Obstruction of the intestinal tract also occasionally occurred during development. Although trisomy 21 is the autosomal trisomy most compatible with survival through the period of gestation, only about a third of affected embryos and fetuses were actually live born [4].

Trisomy 21, the presence of a supernumerary chromosome 21, results in a collection of clinical features commonly known as Down syndrome (DS). DS is among the most genetically complex of the conditions that are compatible with human survival post-term, and the most frequent survivable autosomal aneuploidy. Mouse models of DS, involving trisomy of all or part of human chromosome 21 or orthologous mouse genomic regions, are providing valuable insights into the contribution of triplicated genes or groups of genes to the many clinical manifestations in DS. This Endeavour is challenging, as there are >200 protein-coding genes on chromosome 21 and they can have direct and indirect effects on homeostasis in cells, tissues, organs and systems. Although this complexity poses formidable challenges to understanding the underlying molecular basis for each of the many clinical features of DS, it also provides opportunities for improving understanding of genetic mechanisms underlying the development and function of many cell types, tissues, organs and systems. Since the first description of trisomy 21, we have learned much about intellectual disability and genetic risk factors for congenital heart disease. The lower occurrence of solid tumors in individuals with DS supports the identification of chromosome 21 genes that protect against cancer when overexpressed. The universal occurrence of the histopathology of Alzheimer disease and the high prevalence of dementia in DS are providing insights into the pathology and treatment of Alzheimer disease. Clinical trials to ameliorate intellectual disability in DS signal a new era in which therapeutic interventions based on knowledge of the molecular pathophysiology of DS can now be explored; these efforts provide reasonable hope for the future [5].

The present study aimed to measure the extent of community awareness about Down Syndrome and spread awareness in Hail City in order to better understand the needs of a patient with this syndrome, diagnosis, treatment and to gain support from the community. Moreover, To increase the knowledge and awareness about Down Syndrome in Hail City community, whether Saudi or non- Saudi, by letting them search and think more about it. Thus changing what they thought of false beliefs about this syndrome.

MATERIALS AND METHODS

Material: Adult persons lived in Ha'il region, aged 20 years and more, were the material of the present study. They were asked to answer a previously designed electronic self-administered questionnaire. Ethical informed consent was taken before from each participant

An online questionnaire was designed and distributed to Hail resident via various social media apps The questionnaire measured several variables and was

composed of two sections. The first section covered the sociodemographic of the participants, including age, gender, nationality, residence, educational level. . The second section comprised of ten questions and included informed consent and the questions regarding signs ,symptoms ,complication and management

Statistical Analysis:

Data analysis: Statistical Package of (SPSS v.26) was used to describe the basic features of the data in the present study, through frequencies, percentages and Pearson's Chi-squared test (χ^2). They were used to find the association between the level of awareness (Good-Poor) and socio-demographic variables; Since Pearson's Chi-squared test (χ^2) is a statistical test applied to sets of categorical data to test the independence of two variables, expressed in a contingency table. Independence means that knowing the value of the row variable does not change the probabilities of the column variable (and vice versa). Another way of looking at independence is to say that the row percentages (or column percentages) (remain constant from row to row (or column to column)).

RESULTS

Demographic Results: Table 1 showed that the total sample size was (364), 95.1% were Saudi, while 4.9% were Non-Saudi. 68.1% were females while 31.9% were males. 56% were 20 to 29 years old, while 11.5% were over 50 years old, About educational level, the highest group was (University educated) by 65.7%, while the least was (Uneducated) by 0.5%. As regards regions, 67.3% were from Hail, while 32.7% were from regions other than Hail. Finally about marital status, 51.1% were single and 48.9% were married. (See. Table I)

Table 1: Demographic data of the sample study (n=364)

| Demographic | Groups | Frequency | Percent |
|-------------------|---------------------|-----------|---------|
| Age | 20 to 29 | 204 | 56.0 |
| | 30 to 39 | 64 | 17.6 |
| | 40 to 49 | 54 | 14.8 |
| | Over 50 years old | 42 | 11.5 |
| Gender | Male | 116 | 31.9 |
| | Female | 248 | 68.1 |
| Nationality | Saudi | 346 | 95.1 |
| | Non-Saudi | 18 | 4.9 |
| Educational level | Uneducated | 2 | 0.5 |
| | Primary school | 4 | 1.1 |
| | Intermediate school | 16 | 4.4 |
| | Secondary school | 103 | 28.3 |
| | University educated | 239 | 65.7 |
| Region | Hail | 245 | 67.3 |
| | Other than Hail | 119 | 32.7 |
| Marital status | Married | 178 | 48.9 |
| | Single | 186 | 51.1 |

Awareness Results: Table II showed the participants responded to the ten questions of the awareness of Down syndrome. Total score ranged between 0 to 10. Since poor level considered for the total score (0 to 4) and good level considered for the total score (5 to 10). Results showed that 239 participants out of 364 (65.7%) of the total sample had good awareness while 34.3% of the total sample had poor awareness. (Table II)

Table 2: Respondents' Awareness level of Down syndrome (n=364)

| | | Frequency | Percent |
|---|---|-----------|---------|
| Have you ever heard of Down syndrome or seen someone suffered of it? | Yes* | 324 | 89.0 |
| | No | 40 | 11.0 |
| You think that Down syndrome is: | Disorder that includes repetitive movements or unwanted and uncontrollable symptoms. | 41 | 11.3 |
| | A rare genetic disorder that affects brain development and leads to severe mental and physical disabilities. | 73 | 20.1 |
| | A genetic disorder caused by the presence of a third version or part of chromosome 21.* | 175 | 48.1 |
| | Don't know | 75 | 20.6 |
| Do you think that Down syndrome is a genetic defect? | Yes* | 170 | 46.7 |
| | No | 58 | 15.9 |
| | Maybe | 136 | 37.4 |
| Is it easy to identify it after birth? | Yes* | 207 | 56.9 |
| | No | 40 | 11.0 |
| | Maybe | 117 | 32.1 |
| Do you think there are symptoms/physical signs of this syndrome: | Grunting & coughing | 10 | 2.7 |
| | Prominent tongue, short stature and neck, eyes raised to the top, flat nose* | 256 | 70.3 |
| | Repeating words, using obscene words | 21 | 5.8 |
| | Don't know | 77 | 21.2 |
| In your opinion what is the cause of this syndrome? | Autoimmune causes | 11 | 3.0 |
| | Unknown causes | 44 | 12.1 |
| | Inherited (Genetic) causes* | 221 | 60.7 |
| | Don't know | 88 | 24.2 |
| What are the risk factors that increase the chance of this syndrome? | Frequent use of devices | 42 | 11.5 |
| | The birth of a child with this syndrome increases as the mother ages* | 183 | 50.3 |
| | Sex | 9 | 2.5 |
| | Don't know | 130 | 35.7 |
| If you think there are complications of this syndrome, what are they? | Heart health problems, dementia, leukemia* | 130 | 35.7 |
| | Loss of social relationships | 29 | 8.0 |
| | Hyperactivity disorder, obsessive compulsive disorder, anxiety | 79 | 21.7 |
| | Don't know | 126 | 34.6 |
| What do you think about the cure for this syndrome? | Anti-seizure medications | 27 | 7.4 |
| | Antipsychotic medications | 14 | 3.8 |
| | Early training programs can help improve their skills (speech therapy - physical therapy - educational therapy).* | 246 | 67.6 |
| | Don't know | 77 | 21.2 |
| What are the methods of diagnosis? | Chromosome testing (karyotype test)* | 215 | 59.1 |
| | Magnetic resonance imaging | 23 | 6.3 |
| | Otoscope / Ophthalmoscope | 6 | 1.6 |
| | Don't know | 120 | 33.0 |
| Poor (1 to 4) | | 125 | 34.3 |
| Good (5 to 10) | | 239 | 65.7 |

*: Correct answers which considered as good Awareness

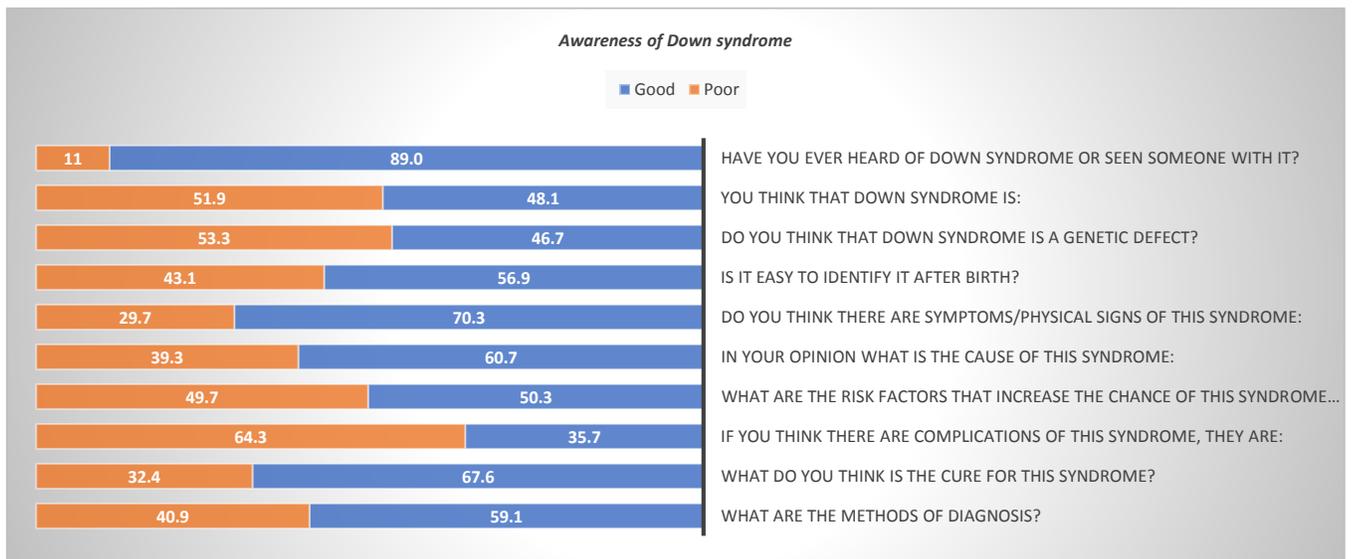


Figure 1: Association between Awareness of Down syndrome and demographic variables

Table III showed the results of Chi square tests for association between awareness of Down syndrome and demographic variables. It indicated that there was statistically significant association ($p < 0.05$) between the level of awareness and gender, age, educational level and

marital status. Whereas , there was no statistical significant association ($p > 0.05$) between the level of awareness and both of nationality and region; due to the convergence of percentages among groups. (**Table III**)

Table 3: Association between Awareness of Down syndrome and demographic variables

| Demographic | Groups | | Level of Awareness | | Chi-Square | P-value |
|-------------------|---------------------|----------------|--------------------|-------|------------|---------|
| | | | Poor | Good | | |
| Age | 20 to 29 | Count | 56 | 148 | 27.547 | 0.000** |
| | | % within Level | 27.5% | 72.5% | | |
| | 30 to 39 | Count | 24 | 40 | | |
| | | % within Level | 37.5% | 62.5% | | |
| | 40 to 49 | Count | 16 | 38 | | |
| | | % within Level | 29.6% | 70.4% | | |
| | Over 50 years old | Count | 29 | 13 | | |
| | | % within Level | 69.0% | 31.0% | | |
| Gender | Male | Count | 75 | 41 | 69.391 | 0.000** |
| | | % within Level | 64.7% | 35.3% | | |
| | Female | Count | 50 | 198 | | |
| | | % within Level | 20.2% | 79.8% | | |
| Nationality | Saudi | Count | 119 | 227 | 0.009 | 0.574 |
| | | % within Level | 34.4% | 65.6% | | |
| | Non-Saudi | Count | 6 | 12 | | |
| | | % within Level | 33.3% | 66.7% | | |
| Educational level | Uneducated | Count | 1 | 1 | 23.351 | 0.000** |
| | | % within Level | 50.0% | 50.0% | | |
| | Primary school | Count | 3 | 1 | | |
| | | % within Level | 75.0% | 25.0% | | |
| | Intermediate school | Count | 5 | 11 | | |
| | | % within Level | 31.3% | 68.8% | | |
| | Secondary school | Count | 53 | 50 | | |
| | | % within Level | 51.5% | 48.5% | | |
| | University | Count | 63 | 176 | | |
| | | % within Level | 26.4% | 73.6% | | |
| Regions | Hail | Count | 84 | 161 | 0.001 | 0.533 |
| | | % within Level | 34.3% | 65.7% | | |
| | Other than Hail | Count | 41 | 78 | | |
| | | % within Level | 34.5% | 65.5% | | |
| Marital status | Married | Count | 75 | 103 | 9.385 | 0.002* |
| | | % within Level | 42.1% | 57.9% | | |
| | Single | Count | 50 | 136 | | |
| | | % within Level | 26.9% | 73.1% | | |

** . Correlation is significant at the 0.01 level (2-tailed).

* . Correlation is significant at the 0.05 level (2-tailed).

DISCUSSION

The present study was conducted to explore the awareness level in general about Down syndrome among people in Hail city. In the present study, total sample size of (364) responded to the questionnaire. 95.1% were Saudi, 68.1% were female while 31.9% were male. The highest percent 56% were 20 to 29 years old, 65.7% had university degree, 67.3% were from Hail, while 32.7% were from regions other than Hail, 51.1% were single and 48.9% were married.

The result of the present study revealed that the overall level of awareness about Down syndrome was moderate (65.7 %). Similar results were previously recorded in studies in Jeddah, KSA in 2018) [6] in Riyadh ,KSA in 2020 [7] cities, with sixties percent with good awareness of DS.

On the other hand, a lower level of knowledge was found in an early study conducted in (Jeddah 2017) [8] where the level of knowledge was only (20 %). The difference of the two results of Jeddah might be due to the sample size was 1253 in (Jeddah 2018)[6] which indicated accurate results, whereas in Jeddah, 2017 [8] , the sample

size was 421 participants which did not represent the whole population of Jeddah city. Therefore, the results could not be generalized.

Comparing the present results (65.7%) with internationally studies, along with the Australian [9], Thai [10], Greek [11] studies, only the Dutch study [12] found that the majority of the participants had sufficient knowledge about DS. However, a lower level of awareness was reported in Australia and the United Kingdom [13].

Many factors helped in explaining the present findings. The results revealed that people who participated in the present study heard about Down syndrome from different sources, since the Saudi Kingdom has a great role to care about DS in many aspects. Also, most of the participants in the present sample were educated (98.45 %), which might explain the present finding. When asking about Down syndrome, it was found that most of participants in the present study (89%) heard about the syndrome by its scientific term (Down syndrome), which was closely similar to the finding of a study conducted in Australia.[9].

In the present work, 89% of the total participants had heard of Down syndrome or seen someone with it. 48.1% thought that Down syndrome is a genetic disorder caused by the presence of a third version or part of chromosome 21. About 46.7% answered that Down syndrome is a genetic defect and 37.4 % answered (maybe). 56.9% reported that it was easy to identify it after birth. A higher percentage of correct answer was reported in (Jeddah 2018) study,(77.7 %) [6]. The reason of the difference was that 37.4 % of our participants answered (maybe), which meant that they needed just a little more awareness to be sure of this information and if we supposed that they answered (yes), the percentage of the correct answer (yes) would be 84.1 %, which was nearly similar to (Jeddah 2018) study's results [6].

In the present study, 70.3% thought that the physical signs of this syndrome were prominent tongue, short stature and neck, eyes raised to the top, flat nose. Also, 60.7% believed that the cause of this syndrome is inherited (Genetic) causes and that the risk factors that increase the chance of this syndrome occurring was the increase of the mother ages (50.3%).. A study conducted in Australia revealed a lower percentage (30%) of their sample who stated the Inherited (Genetic) causes of DS [9] This made our result (60.7 %) reasonable.

As regards the diagnostic methods of Down syndrome, the present results showed that 59.1 % of the participants responded that the method of diagnosis was chromosome testing (karyotype test). A study conducted in Riyadh , in 2020 [7] revealed that 81.1 % of their sample also correctly stated that the methods of diagnosis were (appearance, genetic test), which indicated higher percent than in our study. The reason of the difference between the present result, for this question, and the Riyadh study's result was the difference of number of answer choices on this question. Also the difference in the number of participants. However, in the present work, the least percent of good awareness was 35.7% for the complications of this syndrome.

When asking about the early training programs of Down syndrome, the majority (67.6 %) of the present work's participants responded that the cure for this syndrome was early training programs which could improve their skills (speech therapy - physical therapy - educational therapy), while 11.2 % of the participants responded incorrectly .Many previous researches also revealed the importance of early training programs to improve the quality of life for the patients of DS [6-8, 11]

In the present research, Chi square test results showed that there was significant relation between awareness of Down syndrome and gender ($p < 0.01$); since results showed higher percentage of good awareness about Down syndrome for females with percent 79.8%. About age, there was significant relation between awareness of Down syndrome ($p < 0.01$) and age; since results showed higher percentage of poor awareness for the participants who had over 50 years old with percent 69%. About education level, there was significant relation between awareness of Down syndrome and level of education ($p < 0.01$); since results showed higher percentage of good awareness for the participants who had university degree with percent 73.6%. Marital status also

showed significant relation with awareness of Down syndrome a ($p < 0.05$); since results showed higher percentage of good awareness for single participants with percent 63.4%. Otherwise, there was no statistically significant association ($p > 0.05$) between awareness and both of nationality and region; due to the convergence of percentages among groups. Nearly similar results were found in previous researches [6-9].

CONCLUSION

Down syndrome is a condition in which a person has an extra chromosome, which can cause both mental and physical challenges for the baby. The present study aimed to explore the awareness level in general about Down syndrome among people in Hail city. The Kingdom of Saudi Arabia pays wide attention and care to people with special needs, including Down syndrome patients, as it provides them with centers and schools that offer them training programs to learn skills and crafts that will help them in their future lives. It also provides care and training to their families. The present study found that majority of the respondents were aware of Down Syndrome in KSA (65.7%). Furthermore, good awareness was detected for the participants who had university degree with percent 73.6%, for females with percent 79.8% and for single participants with percent 63.4%. On the other hand, higher percentage of poor awareness was found in old age participants with percent 69%. Also, the participants had poor knowledge about its complications. More Health education for public, online seminars and more research are needed to raise the public awareness and spotlight to patients suffering from this syndrome.

Consent: Ethical informed consent was taken before from each participant .

Ethical Consent Approval: An ethical approval for this study was obtained from ethical committee of the University of Ha'il, number H-2021-187, dated 04\10\2021. Participants in the study were informed that their participation was voluntary, and that their contribution was of great value. No personal identifiers were collected. All authors declare that informed consent was obtained from the participants for publication of this original article.

REFERENCES

1. Mohammad K, Mansoor S, Majid K. Down syndrome: current status, challenges and future perspectives. *International journal of molecular and cellular medicine* 2016; 5(3): 125.
2. Grieco J, Pulsifer M, Seligsohn K, Skotko B, & Schwartz A. Down syndrome: Cognitive and behavioral functioning across the lifespan. In: *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. 2015: 135-149.
3. Jaruratanasirikul S, Kor-Anantakul O, Chowwichian M, Limpitikul W, Dissaneevate P, Intharasangkanawin N, Sattapanyo A, Pathompanitrat S, Sriplung H. A population-based study of prevalence of Down syndrome in Southern Thailand. *World Journal of Pediatrics* 2017; 13(1): 63-69.
4. Epstein CJ. Down syndrome. In: *Abnormal States of Brain and Mind*. Birkhäuser, Boston, MA, 1989 :43-44.
5. Antonarakis S E, Skotko B G, Rafii M S, Strydom A, Pape S E, Bianchi D W, Sherman S L, Reeves R H. Down syndrome. *Nature Reviews Disease Primers*, 2020; 6(1): 1-20.

6. Anwer, F., Alhaddad, M. H., Basonbul, R. A., Butt, N. Sh., Noor, M. I., Malik, A. A. (2018). knowledge and attitude towards down syndrome, *Proceedings SZPGMI journal*, 32 (1), 56-65.
7. Shalabi, A. F., Shrouro, . O. A., Sibaa, . R. H., Alalida, . S. B., Alammr, . N. Z., Almoumen, .F. A., Tamim, . B. K., Natto, . Y. S., Alamri, . R. E. & Abdulaziz, . A. M. (2020) Assessment of awareness level toward Down syndrome in Riyadh, Saudi Arabia. *International Journal of Medicine in Developing Countries*, 4 (2), 423-428.
8. Binjahlan, Y., Binjahlan, M., Alqurashi, A., Alqurashi, G., Zirari, M., Alturkistani, F., Tolah, M., Khedrawi, A. Assessment of Knowledge, Attitude and Practice toward Down Syndrome in Jeddah City, Saudi Arabia 2016. *The Egyptian Journal of Hospital Medicine*, .2017 66(1), 146-151. doi: 10.12816/0034645.
9. Mulvey S, Wallace E. Levels of knowledge of down syndrome and down syndrome testing in Australian women. *Aust N Z J Obstetr Gynaecol*. 2001;41(2):167–171. <https://doi.org/10.1111/j.1479-828X.2001.tb01202.x>
10. Pruksanusak N, Suwanrath C, Kor-Anantakul O, Prasartwanakit V, Leetanaporn R, Suntharasaj T, et al. A survey of the knowledge and attitudes of pregnant Thai women towards Down syndrome screening. *J Obstet Gynaecol Res*. 2009;35(5):876–81. <https://doi.org/10.1111/j.1447-0756.2009.01035.x>
11. Gourounti K, Sandall J. Do pregnant women in Greece make informed choices about antenatal screening for Down's syndrome? A questionnaire survey. *Midwifery*. 2008;24(2):153–162. <https://doi.org/10.1016/j.midw.2006.09.001>
12. van den Berg M, Timmermans DRM, ten Kate LP, van Vugt JMG, van der Wal G. Are pregnant women making informed choices about prenatal screening? *Genet Med*. 2005;7(5):332–338. <https://doi.org/10.1097/01.GIM.0000162876.65555.AB>
13. Chilaka VN, Konje J, Stewart C, Narayan H, Taylor D. Knowledge of down syndrome in pregnant women from different ethnic groups. *Prenat Diagn*. 2001;21(3):159–164. [https://doi.org/10.1002/1097-0223\(200103\)21:3<159::AID-PD20>3.0.CO;2-V](https://doi.org/10.1002/1097-0223(200103)21:3<159::AID-PD20>3.0.CO;2-V)