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Investigation of the lawsuits regarding Down syndrome

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ABSTRACT

Objective: To examine the number of cases filed about Down syndrome in terms of its numbers, causes and consequences, to provide an overview of what doctors should pay attention to when informing and consulting patients and during follow-up and recommend solutions for decreasing the number of malpractice cases. (Discussing the legal aspect of the decisions is beyond the scope of this research.)

Materials and Methods: 'Down', 'Down sendromu' 'Down's, 'trizomi 21 ', 'trisomi 21' and 'trisomy 21' was written to ' https://karararama.yargitay.gov.tr/ ' and 'https://karararama.danistay.gov.tr/' web addresses search engines and the data was examined with Microsoft Excel or with R version 4.0.5 (2021-03-31) for bias and frequency table was used and the results were examined.

Results: A total of 53 cases were found. 49 supreme court and 4 Council of State court decisions are found. The cases are from 27.10.2009 to 13.10.2021.

Conclusion: A total of 39 different Down syndrome cases were examined, as 6 of the 53 cases were related to the same cases and 8 of them were cases not related to Down syndrome. 28 cases are "doctor negligence", 5 are "reckless killing", 1 "material mixing in the genetic center", 1 "unauthorized use of the child's photo", 1 "stealing money from the child", 1 "intentionally injuring the child", 1 'inheritance request for the child' and 1 on 'guardianship'.

Keywords: Down syndrome, Prenatal screening, Prenatal diagnosis, Malpractice, Lawsuit

1. INTRODUCTION

Down syndrome or trisomy 21 (having 3 of the 21st chromosomes) is the most common chromosomal disorder and the most common non-hereditary cause of intellectual disability [1-3]. It is seen in 1/600-800 people. According to the data analysis of the Turkish Statistical Institute (TUIK), the incidence of Down syndrome in the population is 1/530. In addition to intellectual disability, congenital heart anomalies, hypothyroidism, cataracts at an early age, hearing loss, intestinal anomalies are also common [4]. Early dementia and Alzheimer's are important causes that shorten life expectancy. People with Down syndrome have a characteristic facial appearance such as upslanting palpepral fissures, epicanthal folds, small ears and flat nasal bridge (OMIM#190685). Failure of chromosomes to separate during meiosis is the underlying cause of 95% of the cases [4]. The remaining causes include translocations and mosaicism. While the risk is 1/1400 for each live birth at the age of 24, this rate increases to 1/350 at the age of 35 and 1/45 at the age of 43 [5].

Although, advanced maternal age (over 35 years of age) is an important risk factor, giving birth to a baby with Down syndrome is more common in mothers under the age of 35 since most of the pregnancies occur in women under the age of 35. According to Turkish Statistical Institution (TUIK) data, there have been a total of 7,841.668 deliveries in Turkey in the last 5 years. 14% of them (n=1,097,834) are by women over 35 years old and 86% (n=6.743,834) are by women under 35 years old [6].

Double, triple and combined tests and detailed ultrasound examinations are recommended for all pregnant women, regardless of age, in the prenatal screening guidelines of the Ministry of Health in Turkey. These are suggestions only and not mandatory. The 'extracellular free DNA' test, which has a sensitivity of approximately 99% in the detection of Down

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syndrome and called non-invasive prenatal test (NIPT), is not among the recommendations made by the Ministry of Health and is not performed in public hospitals.

According to the Biomedical Contract and the Patient Rights Regulation, there is no need to obtain written consent when ordering these screening tests and since, there is no document of proof, the majority of the cases brought to the court due to the birth with Down syndrome doctors are accused of negligence for not offering these tests to the pregnant women [6]. These accusations may be the result of not informing the patient enough or the patients not understanding the information.

Even in high-risk pregnancies (in our country, if the risk is less than 1/270 for having a baby with Down syndrome, it is considered as high risk), definitive diagnostic tests such as amniocentesis and chorionic villus sampling (CVS), are offered to the patient as a recommendation only and again are not mandatory. CVS is usually done between 10-14 weeks of pregnancy and amniocentesis is done after the 16th week of pregnancy until the 20th week. Since, it is an invasive test, complications such as infection and bleeding may be seen. Death of the fetus and the mother are seen very rarely. Although, consent is obtained from the pregnant woman for these tests (some experts also obtain consent from the father), some of the lawsuits are filed for the crime of "reckless killing" due to the death of the fetus or the pregnant woman [7].

The purpose of this research is to examine the number, reasons and results of the lawsuits filed regarding Down syndrome and to provide a general perspective on what doctors should pay attention to during informing, consulting and follow-up of patients and to recommend solutions for reducing the number of malpractice cases. To the best of my knowledge, this is the first study examining the lawsuits for Down syndrome in Turkey.

2. MATERIALS and METHOD

In this observational study, we examined each of the results for searches on the terms "Down", ""Down sendromu", "Down's", "trisomi 21", "trizomi 21", and "trisomy 21" in the two search engines https://karararama.yargitay.gov.tr/ (supreme court cases) and "https://karararama.danistay.gov.tr/' (state of council cases) .Search on "Down" produced 49 supreme court decisions, and search on "Down sendromu" produced 21 supreme court and 4 state of counsil decisions. Search on "Down's", "trisomi 21", "trizomi 21", and "trisomy 21" produced no results. The dates of the decisions ranged from 27.10.2009 to 13.10. 2021.The focus of this study is to examine the reasons of the court cases, and the court outcomes (Table I).

This study was approved by the Clinical Research Ethics Committee of Demiroglu Bilim University (approval number 44140529).

Statistical Analysis

All statistical evaluations as well as handling descriptive data (e.g., frequency table) throughout this study was carried with Microsoft Excel or with R version 4.0.5 (2021-03-31). To check whether there is any bias in selection of cases with respect to the decision, Chi-square test is used with the null hypothesis that there is no difference between expected number and observed number of cases, therefore there is no bias. P value less than 0.05 was considered as significant.

3. RESULTS

We took into account 39 of the 53 court case results. Of the 14 court case results we did not take into account, 6 were related to the same court cases and 8 were court cases unrelated to Down syndrome (they just happened to have the terms "Down" or "sendrom" in the decision texts; for example one court case was about a rap song that contained the word "Down" in its lyrics, and two cases related to hemolysis, elevated liver enzymes, low platelet count (HELLP) syndrome.

Of the 39 court cases, 71.7 % (28 cases) were for doctor's negligence, 12.8% (5 cases) for manslaughter through negligence, 1 for mixing-up of sample material at the genetic testing center, 1 for unauthorized use of the photograph of a child with Down syndrome in a test book, 1 for stealing money from a child with Down syndrome, 1 for injury with intent of a child with Down syndrome, 1 for demanding the inheritance rights for a child with Down syndrome, and 1 for guardianship (Table I). There were no bias in the selection of cases with respect to the decision (p=0.12).

Table I. *Cases related to Down syndrome and their results that are found in data on 'karararama.yargitay.gov.tr' and 'karararama.danistay.gov.tr' websites*

Cases	Outcome			
w	Acceptance/ Compensation	None/ Dismissal / Acquittal	Appeal	Total and Frequency
Doctor's negligence	4	9	15	28 (71.7%)
Unauthorized use of photos	1			1 (2.58%)
Material mixing		1		1 (2.58%)
Deliberate injury			1	1 (2.58%)
Inheritance	1			1 (2.58%)
Stealing money	1			1 (2.58%)
Reckless killing		2	3	5 (12.8%)
Guardianship		1		1 (2.58%)
Total	7	13	19	39 (100%)

In each of the court cases for; stealing from a child with Down syndrome, injury with intent of a child with Down syndrome, unauthorized use of the photograph of a child with Down syndrome, and demanding the inheritance rights for a child with Down syndrome, the court ruled in favor of the plaintiff.

Manslaughter through negligence

One court case was related to the death of a mother and baby with Down syndrome after birth. One court case was related to the death of the fetus and of the mother from septic shock, following a second amniocentesis after an unsuccessful first

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amniocentesis, two court cases were related to the death of the fetus after an amniocentesis, and one court case was related to the death of a child with Down syndrome the following morning after being sent home with a diagnosis of acute tonsillitis and being prescribed antibiotics. Three of these cases were re-opened on appeal, and in two of them the doctors were acquitted. The outcome of the appeals is not known.

Mixing-up of sample material at the genetic testing center

The parents of a child born with Down syndrome opened this court case with the claim that they did not have an abortion because their amniocentesis test result was normal, and that another set of parents had an abortion because their amniocentesis test showed Down syndrome whereas their baby was normal. It is not known whether the other parents who had an abortion even though their fetus was normal, opened a court case or not. As it was not able to be proven that the samples were mixed up at the genetic testing center, the center was found not guilty.

Doctor's negligence (malpractice)

In these court cases the plaintiffs sued for medical malpractice accusing the doctors of having not sufficiently informed them regarding screening tests, did not get informed consent, did not act correctly regarding the diagnosis, did not recommend advanced testing, were negligent in performing ultrasound, did not analyze the ultrasound results, did not request consultation, and did not perform amniocentesis. Of the 39 court cases, 28 were in this category. In 9 of the cases the doctors were acquitted, and in 4 cases the doctor was ordered to pay compensation. The remaining 15 cases went to appeals, and the result of the appeals are not known.

4. DISCUSSION

Definitive diagnosis of Down syndrome during pregnancy can only be made by invasive diagnostic methods such as CVS or amniocentesis. If the fluorescent in situ hybridization (FISH) and chromosome analysis of the samples taken by these methods show the presence of three chromosomes in the 21st chromosome, Down syndrome diagnosis is made. A meta-analysis study in 2019 showed that amniocentesis caused approximately 0.91% and CVS caused 1.39% pregnancy loss [8]. In the same article, the miscarriage rate in the control group was 1.23% and the authors concluded that the procedure related risks of miscarriage following amniocentesis and CVS are lower than expected. In a study by O'Connor et al, 11.7% of mothers of a fetus with Down syndrome and who continued their pregnancy had miscarriage and 26.4% had stillbirth [9]. If these types of invasive methods are performed by expert teams, the miscarriage rate is very low and maternal death was not reported in the literature. However, complications such as fetal loss, amniotic fluid leakage, chorioamnionitis, needle injury and vaginal bleeding may occur in all of these procedures. During these procedures, evaluation by ultrasound, evaluation in terms of infection risk and Rh prophylaxis should be performed [10].

In this study 12.8% of cases were about complications of the procedures that resulted in either the death of the mother, the fetus or both. This is a high number compared to the expected deadly complication rates of these procedures [8]. In the literature review, we could not find any prevalence data in our country showing the complications of these invasive methods. Studies were published only as single center experiences. There is a need for a multicenter study on this subject in our country. Considering that maternal death occurred in two of the cases in our article, the importance of the subject becomes even more understandable.

Screening tests only show the magnitude of the risk and if this risk is greater than 1/270 in our country, invasive diagnostic methods are recommended for the mother [11]. The social security institution pays the fees for double, triple, quadruple screening tests and second-level ultrasonography performed by radiologists. It also covers the cost of CVS and amniocentesis. It does not pay for the NIPT tests which is being used widely in the developed world [12]. However, studies have shown that NIPT tests reduce the number of invasive tests [13]. At the same time, Huang et al., concluded in their study that the NIPT test screening is the most cost-effective screening test and can increase the effectiveness of prenatal aneuploidy screening by reducing the number of patient visits and providing earlier results [14]. There is a need for research on the effectiveness of NIPT tests for our country, which are currently more costly than double and triple screening tests.

The results of screening tests are reported as 'normal', 'negative' or 'positive' by some laboratories. This situation leads families to reach the wrong conclusion that these tests give definite results. In most of the 'doctor negligence' cases filed, there are accusations that the doctor did not sufficiently inform the patient and did not direct them to invasive diagnostic methods. As written in the guide of the Ministry of Health, pregnant women are informed about prenatal screening and diagnosis, and a screening test is done if requested and written consent is not obtained when ordering or not ordering these tests [11]. However, obtaining written consent does not always end the legal disputes because what is written in the consent may be found insufficient [12]. It is especially important that the obstetrician takes time to explain these tests to the family in detail and make sure that they understand. If possible, the obstetrician's directing the family to a medical geneticist before invasive diagnosis will help the family to make a more informed decision and reduce possible lawsuits. At the same time, the use of more reliable NIPT tests will reduce the use of invasive diagnostic tests and ultimately reduce the number of cases. These tests should be easily accessible to everyone, and these tests should be carried out in our country (most of the tests are sent abroad).

If the pregnant woman does not want a definitive diagnosis, a letter of rejection should be obtained. If she wants a definitive diagnosis, she is directed to the physician who will perform amniocentesis or other invasive diagnostic methods. The obstetrician should inform the pregnant woman in terms of the amniocentesis procedure and obtain written consent. To perform genetic analysis from the amniotic fluid sample, the

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genetic diagnosis center must obtain written consent also. It is also helpful to get written consent from the baby's father.

On another note, while talking about prenatal screening and diagnostic tests; it should also be explained that these tests do not make a diagnosis for all diseases. Diagnostic tests only look for trisomy 21, trisomy 13, and trisomy 18 syndromes unless a different test is requested.

The limitations of this study are there are not many cases that are present in the supreme court and state of council web sites. Also, the results of most of the appeals are not known. These websites should be improved by adding more cases and the results of these cases.

As a result, false positive and false negative results can be reduced by using screening tests with high sensitivity and specificity. It should also be explained that prenatal screening and diagnostic tests are not informative for all diseases.

During patient consultation, care must be taken that the consult is not directive, psychological support should be provided to the parents after the diagnosis, the decision to continue pregnancy should be left to the parents and the decision should not be judged [15]. Efforts should be made to reduce the number of patients per doctor so that doctors can give patients the required time. Written consents should be taken, and doctors must make sure that the patients understand what is written on the consent form. All these measures can improve the doctor-patient relationship, enable healthier decisions, and reduce the number of lawsuits.

Compliance with the Ethical Standards

Ethical Approval: Ethical approval for this study was obtained from Clinical Research Ethical Committee of Demiroglu Bilim University (approval number 44140529)

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Author Contribution: HK: Designed, searched the cases in the websites , did literature review and wrote the manuscript.

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