Experience of mothers with Down syndrome children at the time of diagnosis

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ABSTRACT

Introduction. Down's syndrome (DS) is a common genetic disorder. The families of children with DS generally do not receive sufficient information at the time of the diagnosis. They are dissatisfied with the manner of healthcare professionals. The objective was to evaluate the experiences of mothers of children with DS at the time of the diagnosis and the communicative attitudes of healthcare professionals.

Population and methods. Mothers who had children with Down syndrome were included. The experiences of the mothers at the time of diagnosis and the attitudes of the healthcare professionals were evaluated by a semi-structured interview.

Results. The study sample was 43 mothers. Eight children had been diagnosed with DS prenatally, and 35 had been diagnosed postnatally. Eighteen of the mothers had received the diagnosis in a pediatric clinic, and 16 had been told of the diagnosis in an obstetric clinic. More than half the mothers had received the first information about their child's diagnosis from a pediatrician. Only five of the 43 mothers had received detailed information. Twenty-three of 32 mothers said that they were met with a negative attitude. Generally, the time put aside to inform the families about the diagnosis was less than 5 min.

Conclusions. Mothers of DS children want more information about DS from healthcare professionals. Furthermore, they want this information to be delivered in a supportive and sensitive manner. Healthcare professionals, especially pediatricians need to allocate sufficient time to discuss DS and its challenges with the family.

Keywords: attitude, diagnosis, Down's syndrome, mothers, pediatrician.

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In countries where genetic counseling is not common, it is usually the pediatrician who first explains the diagnosis to the families.⁴

Having an unhealthy infant, the fear of failing to meet the child's needs, and worries about the future of the child make the time of the diagnosis particularly difficult for families.⁵ The place in where the diagnosis is delivered and the attitudes of the healthcare professionals who deliver the diagnosis are very important in terms of the reactions of the families when they are first informed about the diagnosis. Ideally, the healthcare professional should have a supportive and sensitive manner, give adequate and clear answers to any questions asked by the families, and provide anticipatory guidance services.⁶⁻⁹ The parents' experiences at the time of the diagnosis are extremely important in terms of the subsequent parent-child relationship.¹⁰ Previous studies showed that parents of DS children who established a good relationship with healthcare professionals were more easily attached to their newborns and showed a more positive attitude toward the infant's care.^{7,8,10}

In a study that focused on the healthcare profession worldwide, Skotko⁴ reported that healthcare professionals were unable to provide adequate information in an appropriately sensitive manner when delivering a diagnosis of DS. Even the most experienced doctors accepted that they received insufficient education on how a DS diagnosis should be given in a sensitive manner.¹¹ In a recent study by Dogan et al., it was reported that pediatric residents had a high level of discomfort when communicating with parents of newborn with DS.¹²

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INTRODUCTION

Down's syndrome (DS) is one of the most common genetic syndromes encountered by pediatricians.¹ The diagnosis is generally delivered by pediatricians and genetic specialists to families in the postpartum period.^{2,3}

Although studies on the ideal timing, setting, and type of information to be given to families of DS children have been conducted in Europe and the U.S.¹³⁻¹⁵ there are few such studies in Eastern countries.^{16,17}

In Western cultures, families' expectations may differ from healthcare professionals for this reason. The objective of this study was to evaluate the experiences of mothers of children with DS at the time of the diagnosis and the communicative attitudes of healthcare professionals in Turkey.

POPULATION AND METHODS

This was a descriptive study of mothers of DS children aged less than 6 years who attended the Developmental Pediatrics Department (DPD) of Inonu University Turgut Ozal Medical Center and agreed to participate in the study. The center is the biggest hospital in eastern Turkey. The pediatric department serves approximately 100,000 outpatients annually. The mothers who participated in the study were informed in detail about the scope and aim of the study, and all provided informed consent. The survey was completed over a 5-month period in 2016.

The experiences of all the mothers were evaluated using the "Interview Form for Mothers of Children with Down Syndrome" developed by the DPD (see *Appendix*). The form was developed based on studies published by Sheets¹³ in 2011 and Skotko⁴ in 2009 after obtaining permission from the authors. The form contained 27 questions and included quantitative and qualitative data from yes/no questions and open-ended questions. The form was composed of three sections. The first section focused on demographic information, and the second section focused on the experiences of the families at the time of the diagnosis. In this section, the respondents were asked for their opinions on the most appropriate way to explain a diagnosis of DS. The third section was for the mothers to provide recommendations to healthcare professionals regarding what information should be given to parents immediately after delivering the diagnosis of DS. The participants' answers to the open-ended questions were read and evaluated independently by the researchers. Each interview lasted about 30 min.

This study received ethical approval from the scientific research and publication ethics committee (date: 1.12.2015; decision number: 2015.10-9).

Statistical analysis

Statistical evaluation of the data was performed using the Statistical Package for Social Sciences, version 17 software. Descriptive statistical analysis (frequency, mean/median, and distributions) of the mothers in the sample was performed.

RESULTS

Fifty mothers were invited to take part in the interview. Seven refused, giving a total of 43 mothers in the study. The average age of the mothers at the time of the birth of the DS child was 34.4 ± 5.7 years. *Table 1* shows detailed demographic information.

Eight mothers reported that they received the diagnosis in the prenatal period, and the other 35 (81.4 %) mothers received the diagnosis in the postnatal period. Although 11 mothers stated that the time of receiving the diagnosis was not important, 18 mothers stated that the time of the diagnosis should be in the prenatal period. Seven mothers said that it should be right after the birth, and seven mothers said that it should be after the karyotype analysis.

In terms of where the mothers received the diagnosis, the locations were as follows: neonatal services or pediatric outpatient clinics ($n=18$), obstetrics and gynecology services or pregnancy outpatient clinics ($n=16$), genetic outpatient clinics ($n=5$), doctors' rooms ($n=2$), and delivery rooms ($n=1$). One mother received the diagnosis at home. When they were asked about the most appropriate place to receive the diagnosis,

TABLE 1: Socio-demographic characteristics of the children and the mothers

Sociodemographic characteristics	N = 43	(%)
Gender of the child		
Female	25	(58.1)
Male	18	(41.9)
Child's age (month)		
Mean $\pm$ SD	23.72 ± 20.09	
Minimum-Maximum	1-80	
Mother's age at the childbirth (year)		
< 25	4	(9.3)
25-35	17	(39.5)
> 35	22	(51.2)
Mother's educational level		
Illiterate	3	(7)
< 8 years	25	(58)
≥ 8 years	15	(35)

SD: Standard deviation.

eight cited an obstetrics and gynecology service or pregnancy outpatient clinic, seven cited a pediatric outpatient clinic, five cited the private rooms of a doctor, and three cited genetics departments.

Twenty-three mothers stated that a pediatrician informed them about the diagnosis. In 11 cases, the mothers were informed by gynecologists. Nine mothers were informed by other healthcare professionals.

In terms of the durations of the interviews conducted to inform the family about the diagnosis, the duration was 5 min or less in 25 cases. *Table 2* shows the opinions of the mothers on the kind of information that should be provided about the diagnosis and the types of healthcare attitudes.

In the last section on recommendations to healthcare professionals, the mothers expressed more than one opinion. Of 35 mothers, 18 stated that healthcare professionals should be welcoming (e.g., smile) and understanding (e.g., show empathy and caring) and that they should use appropriate language. Thirteen mothers stated that they should provide sufficient information. Eleven mothers stated that the healthcare professional should appear relaxed and use positive expressions (i.e., optimistic rather than pessimistic).

DISCUSSION

In this study, the mothers of DS children felt that they received insufficient information from healthcare professionals and that the meetings with the healthcare professionals were very short.

Furthermore, they were dissatisfied with the attitudes shown by the healthcare professionals when delivering the diagnosis, which was a time of great shock and sadness for the parents.

Similar to the findings of the present study, Skotko² reported that 87 % of mothers in the U.S. with a DS child received the diagnosis in the postnatal period. Previous studies found that mothers usually wished to be made aware of the diagnosis as soon as the doctors expressed suspicion, even if the diagnosis was uncertain.^{18,19} In cases where the health of the mother was not good or she had not fully recovered after the birth, it was recommended for the doctors to wait until a more appropriate time.^{2,20} In this study, almost half the mothers stated that they would have preferred to have received the diagnosis in the prenatal period rather than after the delivery to allow them to prepare themselves.

Most studies stated that the ideal location to deliver the diagnosis was in a private room, with a single healthcare professional in a one-to-one meeting with the parents of the child.^{4,6} In the present study, most of the families did not receive the diagnosis in a private room.

Some studies suggested that families with DS children felt that both a pediatrician and gynecologist should be present at the initial meeting where the diagnosis is delivered for the first time.^{2,18} In the present study, a pediatrician most commonly informed the parents about the diagnosis, followed by a gynecologist.

In previous studies on DS, no information was provided on the time spent by healthcare professionals with parents at the time of the

TABLE 2. *Open-ended questions (content of information and attitude)*

Questions*	Responses (number of respondents)
What information was given to you? (43 mothers replied)	- The development will be delayed, the infant will be disabled, will not be like normal children, and have genetic difference: 18 - Just the infant has DS: 10 - He/she should get special education: 8 - The infant will have additional health problems: 8 - Detailed information was given: 5 - He/she can die early: 2
How was it told? (How was the attitude of the person giving the information?) (32 mothers replied)	- Explained with a negative attitude (Without detail, only the diagnosis was told, the negative aspects were explained, the information was explained quickly, the medical words were used): 23 - Told with an appropriate expression: 7 - I can not remember how it was told: 2

DS: Down syndrome.

*Mothers expressed more than one opinion.

diagnosis. In the present study, in more than half the cases, the time put aside to inform the parents of the diagnosis was 5 min or less.

Some authors reported the discomfort felt by families when they were given incorrect, out-of-date, and difficult-to-understand information at the time of receiving the diagnosis.⁴ In addition, parents did not welcome the provision of an excessive amount of information about possible health-related conditions, such as leukemia, Alzheimer's disease, and obesity, in the future.^{4,19,20} However, they did require information on the nature of DS and its causes,^{18,21} in addition to information on family support groups and DS communities.^{2,18,21,22} Furthermore, they desired up-to-date information on realistic opportunities available to people with DS today^{2,4,19,21,22} and information on emergency and primary health conditions.^{6,18,21} In the present study, 11 % of the mothers stated that they were informed in detail, whereas this rate varied between 11 % and 84 % in the literature.^{4,20,21} In most cases, after informing the parents about the diagnosis, they were told about some characteristics of DS, most of which was negative. The mothers wanted more detailed information and answers to their questions about future concerns. These questions included issues relating to education, additional health problems, and positive aspects of DS, not only negative aspects.

Many studies have emphasized the importance of doctors having a sensitive and appropriate manner when informing parents about a diagnosis of DS.^{4,8,9,23} Unemotional, rude, and careless doctors caused distress among mothers. In this study, approximately three-quarters of the mothers stated that they were uncomfortable with the negative attitudes of the doctor at the time of the diagnosis, with the remaining mothers stating that the diagnosis was given in an appropriate manner. In terms of expectations regarding healthcare provider attitudes at the time of the diagnosis, the majority of the mothers stated that the doctor should exhibit empathy and appear understanding and comforting.

A previous study reported that mothers who were informed about the positive aspects of DS were more optimistic and positive about the diagnosis and that this increased the attachment between the family and child. In addition, it had a positive impact on the growth and development of the child.²⁴ In a study conducted in Korea, families who were given up-to-date information

about DS and family support groups showed more positive emotions.⁵ In this study, almost all the mothers stated that the diagnosis gave rise to negative emotional reactions, such as sadness, crying, shock, and confusion. The latter may be linked to the provision of insufficient information and to healthcare professions focusing only on negative aspects of DS.

Strengths and limitations of the study

The majority of previous studies were conducted in Western countries. To the best of our knowledge, there are only two studies with similar content conducted in Egypt and Pakistan.^{16,17} In Turkey, there is only one study on the experiences of 11 mothers, whose infants were diagnosed with DS after amniocentesis in the prenatal period. In the study, the mothers' experiences, feelings, and relationships with health professionals at the time of the diagnosis were assessed by a semi-structured questionnaire.²⁵

The limitations of the present study were the small sample size and the fact that the study was conducted in a single center. The use of open-ended questions in the interviews caused difficulties in both the evaluation and discussion. The low rate of response to the questions about the most appropriate way to give information about the diagnosis is likely due to the respondents' lack of knowledge about DS and their low educational levels (65 % of the mothers in the sample had less than 8 years of education). Retrospective data collected from parents can also be subject to recall bias. However, as reported elsewhere,²⁶ 20 years post diagnosis, the parents of DS children recalled the first words uttered at the time of the diagnosis.

CONCLUSIONS

The results of this study showed that mothers of DS children want more information about DS from healthcare professionals. Furthermore, they want this information to be delivered in a supportive and sensitive manner. Healthcare professionals, especially pediatricians need to allocate sufficient time to discuss DS and its challenges with the family. ■

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APPENDIX:
Interview Form for Mothers of Children with Down Syndrome

A. Demographic information

1. Name of the child: Gender:
2. What is your name (Mother's name):
3. How old are you?
4. How old were you at the time of your child's birth?
5. Child's birthday / age:
6. Do you have any children with other conditions [mental or physical problems]?
7. Mother's educational level?

B. The experiences of the mothers at the time of diagnosis

8. When did you receive the diagnosis? Prenatal/postnatal
When is the best time to share the news?
9. Where did you receive the diagnosis?
Where is the best place or setting to deliver the news?
10. Who deliver you the information? Doctor? Nurse? Other professional?
Who is the best person to communicate the news?
11. What information was given to you?
What information should be delivered?
12. How was it told? (How was the attitude of the person giving the information?)
How should the news be communicated?
13. Who was with you?
14. Was your baby with you? Yes/No
15. Did they give you any printed information? Yes/No
16. How long was the interview?
17. When you received the diagnosis, how did it make you feel?
18. What did you do afterwards?
19. Who did you tell?
20. Did you know anyone with Down syndrome before then? Yes/No
21. Did you know much about Down syndrome? Yes/No
22. At the time, what did you believe caused the condition?
23. What do you now believe caused the condition?
24. What are your hopes and dreams for your child?

C. The recommendations

25. Is there anything else you'd like to share or that I didn't ask that is important to you or your family?
  26. What advice would you like to give health care professionals when they diagnose a child with Down syndrome?
  27. What advice would you like to give families that have recently had a child diagnosed with Down syndrome?
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