

Assessment of quality of counselling for down syndrome in Sudan

Abstract

Aim: The aim of the study was to assess the quality of the counselling for the Down syndrome that the parents received at the time of the diagnosis.

Method: Parents of infants with a diagnosis of Down syndrome were interviewed about the counselling received at the time of the diagnosis.

Results: During the study period 109 infants with a diagnosis of Down syndrome were seen.

40 out of the 109 parents (36.7%) did not receive any counselling for Down syndrome, 69(63.3%) were counselled about Down syndrome but 22 out of them (33.9%) felt that the counselling was not good enough.

Conclusion: The training of doctors in counselling and in breaking bad news need to be improved.

Keywords: Sudan, down syndrome (DS), counselling

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Introduction

Down syndrome (DS) is the most common chromosomal malformation affecting about 1:700 live born. The American College of Obstetrics and Genecology (ACOG) and the American College of Medical Genetics now recommends that all pregnant women, regardless of age, be offered prenatal testing for DS.¹⁻³ Although this guidelines exist in the western world but more than 85% of mothers who have children with DS, first received the diagnosis postnatal.^{4,5} We do not have figures for the Sudan, but from our own experience more than 95% receive the diagnosis postnatally. Breaking bad news is one of the most important areas that are facing doctors on regular basis and it is one of the most difficult areas for the doctors to deal with, but unfortunately doctors do not receive adequate formal training in breaking bad news. Bad news may be defined as “any information which adversely and seriously affects an individual’s view of his or her future.”^{6,7} Communicating bad news can be very difficult to the informer even to the most experienced physician, and can be devastating experience to the parents, this experience could influence the way they react or accept the child illness in the long run. Skotko had shown “that the parents reported that being frightened or anxious after learning the diagnosis, and very few rated the overall experience as a positive one. Mothers reported that their physicians talked little about the positive aspects of DS and rarely provided enough up-to-date printed materials or telephone numbers of other parents with children with DS.”⁵ The same views were shared by Steven Ralston who wrote “In general, what I was taught in medical school and in my training is that disability—no matter what its form—is a bad thing and to be avoided at all costs. Lectures or seminars on DS or other genetic syndromes were geared toward the description of the abnormalities... that children with congenital diseases may find their lives to be rich

and valuable was hardly recognized, much less stressed.”⁸ Reports from different parts of the world e.g. UK, Sweden, Australia, USA have reported strong parents dissatisfaction with the way in which the diagnosis was conveyed to them during the immediate postnatal period and also the support provided to them during the same period.⁵ There are good clinical practice guidelines for the delivering of bad news and for the management of children with DS see Table One and Two.^{6,7,9-16} We do not have enough data about the situation in Sudan, but from our personal experience we have noticed that the counselling of parents of DS was not up to the expected standard.

Objective

The objective of this study was to determine if parents received any counselling for Down syndrome at the time of the diagnosis and to assess the quality of the counselling received.

Methods

Newly diagnosed infants with DS who were referred to the Paediatric cardiology clinic for echocardiography were invited to participate in the study. The paediatric cardiology department in Ahmed Gasim Children Hospital covers Khartoum State and receives referral from all parts of Sudan. The study period was six months; we have limited the study to infants to make sure that the experience is fresh in the parent’s mind, so they could remember the details of the counselling. Ethical approval was obtained from the hospital ethical approval committee. A verbal consent was taken from the parents, parents were interviewed by OH and a pre-tested questionnaire was filled. The statistical analysis of this study was performed by utilizing statistical software, Statistical Package for Social Sciences (SPSS) for Windows 17.0 (Table 1 & Table 2).¹⁰

Table 1 Practice guidelines for communicating a prenatal or postnatal diagnosis of Down Syndrome: recommendations of the national society of genetic counselors¹⁰

- a. Tell the parents about the diagnosis as soon as possible, even if the diagnosis is suspected but not yet confirmed. If the diagnosis has not been confirmed by karyotype, explain what physical features or medical concerns are suggestive of the diagnosis.
- b. Ideally, the diagnosis should be delivered in person, by a healthcare professional with sufficient knowledge of the condition. Health care providers should coordinate the message to ensure consistency in the information provided to the family.
- c. Whenever possible, meet with both parents together.
- d. The family should be informed of the diagnosis in their preferred language. If possible, a professional medical interpreter should be present at the time of disclosure.
- e. Discuss the diagnosis in a private, comfortable setting, free from interruptions. Allow time for questions and make plans for a follow-up conversation.
- f. Parents should be provided with accurate and up-to-date information. Information should be given with a balanced perspective, including both positive aspects and challenges related to Down syndrome.
- g. Provide the information in a sensitive and caring, yet confident and straightforward manner, using understandable language that is clear and concise.
- h. Use neutral language and avoid using value judgments when starting the conversation, such as "I'm sorry" or "Unfortunately, I have bad news".
- i. Use sensitive language and avoid outdated or offensive terminology. In the newborn setting, the baby should be present, and should be referred to by name. Use person-centric language, emphasizing that this is a baby who has Down syndrome, rather than a "Downs baby" or a "Down syndrome child."
- j. Allow time for silence tears. Do not feel that you need to talk to "fill the silence". Offer the family time alone.
- k. Assess the emotional reactions of the parents, and validate these feelings. Use active listening and empathic responses to support the parents.
- l. Informational resources should be provided, including contact information for local and national support groups, up-to-date printed information or fact sheets, and books. The opportunity to meet with families who are raising a child with Down syndrome, when appropriate, referrals to other specialist may also be helpful e.g. cardiologist.

Table 2 Essential information for the initial discussion of a diagnosis of down syndrome¹⁰

- a. Down syndrome (DS) is caused by extra genetic material from chromosome 21. DS may be suspected based on physical findings, but the diagnosis is confirmed by chromosome analysis.
- b. Individuals with DS have a variable range of intellectual disability from mild to moderate.
- c. Babies with DS have delays in achieving developmental milestones and benefit from early intervention including physical, occupational and speech therapy.
- d. 80% of babies with DS will have hypotonia.
- e. 50% of babies with DS have one or more congenital abnormality: 40–60% of babies with DS have a heart defect and 12% have a gastrointestinal defect that may require surgery. Assistance with referrals to specialists is appropriate for identified complications.
- f. Children with DS are more like other children than they are different. Raising a child with DS may involve more time commitment than typical children.
- g. Individuals with DS can participate in community sports, activities, and leagues.
- h. Individuals with DS can learn in a special education class or may be included in regular classes and most can complete high school.
- i. Individuals with DS can be employed competitively or in a workshop setting.
- j. Individuals with DS can live independently or in a group home.
- k. Individuals with DS have friends and intimate relationships.
- l. Life expectancy extends into the 50s or 60s.
- m. Information on local support groups, advocacy organizations, early intervention centers, printed material, fact sheets, books, and specialist referral as needed, and the option to contact a family raising a child with DS should be offered.
- n. A personalized recurrence risk for future pregnancies should be offered. We found that in less than one percent of cases the diagnosis was made antenatally, this is in comparison to studies in the west where the percentage was around 15%.^{4,5} This could be explained by the poor antenatal care, absence of screening program and the scarce availability of ultrasound and of highly trained sonographers in the country.

Results

During the study period, 109 infants with a diagnosis of DS were seen. 56 infants were females and 53 were males. In only one case

the possibility of DS was raised antenatally. 40 out of the 109 parents (36.7%) did not know that their children had been diagnosed or suspected to have DS and they had no counselling about DS, although they were referred to the paediatric cardiology clinic with a diagnosis

of DS stated on the echocardiography referral form. 69 (63.3%) knew about the diagnosis/ possibility of DS and were counselled about DS. But 22 out of them (33.9%) felt that the counselling was not good enough, as it did not address their concerns and anxiety.

Discussion

This study showed that more than one third of parents did not know that their child was diagnosed or suspected of having DS. Out of the two third who were told about the diagnosis of DS, one third of them was not happy or satisfied with their counselling. Previous studies tried to address the information that the counsellor should provide to the parents, and how best this should be given.^{4,5,9,12,17,19} Most studies were surveys of parents of DS children who were asked to reflect on the way their healthcare providers delivered the diagnosis of DS. A systemic review by Gysel et al.,²⁰ of communication skills training advised that training in communication skills should be offered to medical and nursing students and to senior medical professionals as well. Study by Sheets et al.,¹² showed that “parents appreciate information about the abilities and potential of people with Down syndrome, as opposed to clinical details. Balancing clinical information with other aspects of the condition, as well as a better understanding of the information parents consider most important, may enable healthcare professionals to more effectively satisfy families’ informational needs following a new diagnosis of DS.” A study from Egypt showed that mother’s likes to be told early, to be told of others with a similar condition, and to be informed of the prognosis.¹⁷ The conversation must take place with both parents in a quiet setting as soon as the diagnosis of DS is suspected. The timing of the disclosure of specific DS related problems must be balanced with respect for the opportunity for parents to welcome their child.¹⁶

Training program for doctors in counselling of DS have been shown to increase the knowledge of doctors and in decreasing the level of discomfort felt by them during the counselling process.¹⁸ There are good clinical practice guidelines for the delivering of bad news and for the management of children with DS.^{6,7,9-16} The use of such protocols should be encouraged as it make it easier for doctors to counsel the parents and this in return should help in providing the best care to the children.

Conclusion

The parents felt that the quality of counselling that they received was not good and even some parents did not receive any counselling at all. Counselling of parents for DS need to be improved, this issue should be addressed by issuing local guidelines for the care of children with DS and by formal training to paediatrician in counselling skills and in delivering bad news. Counselling of parents who have DS children is very important for the long term care of the children, so it must be done in a sensitive and a compassionate manner.

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Conflict of interest

The authors declared there is no conflict of interest.

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