Title: Global survey on obstetricians' perceptions of counselling and decision-making in pregnancies with high risk or confirmed trisomy 21

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INTRODUCTION

The communication of an antenatal diagnosis of Down syndrome (DS) in pregnant women is an experience that leaves a mark on the mother's memory, so the way in which this news is received will greatly influence the attitude and acceptance of the parents (1,2). This information is important for establishing a secure and consistent attachment, as well as for facilitating decision-making about continuing or terminating the pregnancy (3). Survival of infants with DS has improved because of better care (especially of cardiovascular malformations) and survival into mid or late adult life is now expected (4).

Studies of DS have shown increasing trends in affected pregnancies in various parts of the world attributed to increasing maternal age (5). This contrasts with the decrease in births with this syndrome, especially in high-income countries (6). Various screening strategies have been introduced over the years with the aim of offering prospective parents the ability to make informed decisions (7).

Irving et al reported in the UK in a retrospective review of 20 years (2008), that DS affected 1188 pregnancies which either reached 24 weeks gestation or were terminated for fetal abnormality. The prevalence increased from 1.3 to 2.5 per 1000 total births. An antenatal diagnosis was made in 477/1188 cases (40%) and was followed by termination of pregnancy in 389 (81%), stillbirth in 17 (4%) and live birth in 71 (15%). The decision to terminate the pregnancy after antenatal diagnosis varied with maternal age (5).

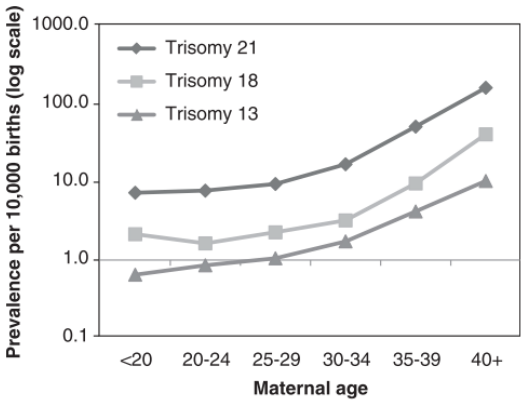

Figure 1 Total corrected prevalence per 10 000 births (log scale) of trisomies 21, 18 and 13 by maternal age, 1990–2009, 12 European countries.

Figure 1: From Loane M et al. Twenty-year trends in the prevalence of Down syndrome and other trisomies in Europe: Impact of maternal age and prenatal screening. Eur J Hum Genet. 2013;21(1):27–33.

Births of children with DS have experienced a sharp decline in the last quarter of a century in Spain, despite the increase in the average age of motherhood (32 years) (8). In countries like Chile, the incidence of births of newborns with DS is higher (2.96/1,000) than in Spain (0.6/1,000), which is interrelated with the possibility of voluntarily terminating the pregnancy (9). For the US, the live birth prevalence for DS in the most recent years

(2006–2010) was estimated at approximately 12.6 per 10,000 with a total of around 5,300 births annually (10).

The global and regional analysis of the burden and trends of Down syndrome (DS) between 1990 and 2019 reveals a complex and heterogeneous reality, marked by advances in public health, socioeconomic differences, and demographic changes (11). The data show that, despite the stability in the global incidence (around 1.2 cases per 100,000 inhabitants), the prevalence has increased, especially in regions with lower sociodemographic development (low and lower-middle SDI). This phenomenon can be explained by the increase in life expectancy and improvements in medical care, which allow more people with DS to survive into adulthood.

The significant decrease in disability-adjusted life years (DALYs) and DS-related mortality worldwide is one of the most positive findings. This trend suggests that medical interventions, access to cardiac surgery, and comprehensive care have had a real impact on the quality and length of life of people with DS, especially in countries with high SDI. However, mortality and the burden of disability remain high in less developed countries, where access to health and social services is limited. Regional differences are notable: countries like Malta, Brunei, and Ireland have the highest prevalence rates, which may be related to restrictive legislation on abortion and older maternal age (11,12). Conversely, the greatest burden of disability and mortality is observed in countries like Haiti, Paraguay, and Algeria, reflecting inequalities in access to diagnosis, treatment, and social support. A relevant finding is the positive correlation between Down syndrome and incidence/prevalence, but a negative correlation with disability and mortality. That is, in more developed countries, more children are born with Down syndrome (probably due to older maternal age and better diagnosis), but they survive more often and with a better quality of life. In contrast, in less developed countries, although the incidence may be lower, survival and quality of life are worse. Finally, the table shows that public health policies, access to prenatal diagnosis, maternal age, and socioeconomic and cultural factors are key determinants in the epidemiology of Down syndrome. These results underscore the need to adapt intervention and support strategies to local realities, prioritizing equity in access to services and improving the quality of life for people with Down syndrome and their families.

	Analysed period	Estimated annual births	Rate (per 10,000 births)	Reduction for prenatal testing
Europe	2011-2015	8.031	10.1	54%
USA	2018	~5000	13.2	37%
Australia	2016-2020	265	8.6	66%
New Zeeland	2016-2020	41	6.9	71%

Table 1: Population trends of Down syndrome by region

Van Casteren et al (2025) described live birth (LB) "reduction percentage" as the percentage of babies with DS that were not born out of the potential number of babies

with DS that could have been born, absent elective terminations. In recent years, these percentages have been high: 37% in the United States, 38% in Eastern Europe, 51% in Northern Europe, 54% in Canada, 59% in Western Europe, 66% in Australia, 71% in New Zealand, and 72% in Southern Europe. Spain tops the list at 83%. The availability and public funding of prenatal screening (especially serum screening, which includes cfDNA) had the greatest impact on reducing births with Down syndrome. The shift from "no screening" to "serum screening available and funded" more than tripled the reduction (OR=3.24, p=0.0001). Maternal age and per capita GNI were also associated with greater reductions, although with smaller effects than screening. Variables such as religiosity or the social justification of abortion did not show significant effects in the model (13).

In DS, prevalence and life expectancy vary by region, influenced by factors such as maternal age and access to prenatal diagnosis. In Latin America and the Caribbean, the birth rate is higher and the reduction due to selective terminations of pregnancy is minimal, while in Europe and the United States, prenatal screening has reduced births with Down syndrome by more than 37% and 50%, respectively (6). Currently, life expectancy exceeds 48 years in Latin America, 58 in Europe, and 54 in the United States (14,15).

In our setting, births of babies with DS have decreased, as in many other European countries (13). Between 1980 and 1985, before the decriminalization of voluntary termination of pregnancy (VTP) (1985), the total birth rate was 14.7 per 10,000 newborns. Between 1986 and 2022, it was 8.86 per 10,000 newborns. And in 2011, it was 4.8 per 10,000 newborns (16). This reduction has been associated with the widespread implementation of prenatal diagnostic techniques and the availability of VTP, which have become part of routine antenatal care (17). However, the incorporation of prenatal diagnosis into clinical practice should not be interpreted solely to facilitate pregnancy termination. Rather, its primary aim is the early identification of fetal conditions, which allows parents time to understand the diagnosis, process its emotional impact, and prepare appropriately for the child's arrival. Such anticipation facilitates, when indicated, a multidisciplinary approach, as well as the planning of the most appropriate setting for delivery and specialized neonatal care (18–21).

Among the antenatal variables that influence parents' decision-making during pregnancy after prenatal diagnosis of DS is the way in which the news is received by healthcare professionals (22,23). Lack of sensitivity or knowledge, and negative projections about the child's future can make it difficult for parents to accept the diagnosis and reduce their trust in health services. Insensitive and pessimistic communication of a trisomy 21 diagnosis at birth further distresses caregivers (24). Barr et al state that parents report they have been not sufficiently informed and wanted individualized discussion with health professionals (25). In fact, Van Riper et al highlight that healthcare providers are using a communication approach with parents similar to that used decades ago, when negative consequences were thought to be inevitable after the birth of a child with DS (26).

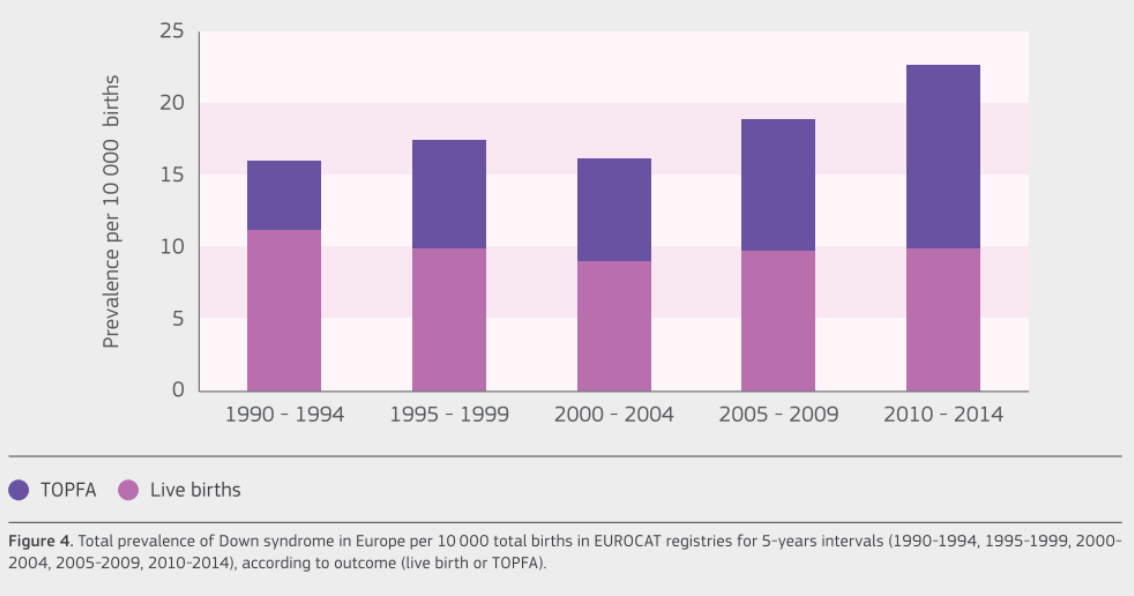

Figure 4. Total prevalence of Down syndrome in Europe per 10 000 total births in EUROCAT registries for 5-years intervals (1990-1994, 1995-1999, 2000-2004, 2005-2009, 2010-2014), according to outcome (live birth or TOPFA).

Figura 2: From Lanzoni M et al. EUROCAT-Surveillance of congenital anomalies in Europe: epidemiology of Down syndrome 1990-2014. 2019 Spell out FOPFA in the figure descriptor.

There are several factors that limit the offering of adequate information to pregnant women who have a probable or certain diagnosis of DS. Healthcare staff training to communicate an antenatal diagnosis of DS to parents is often limited. Skotko et al. show that opportunities for learning on how to communicate a diagnosis of DS or to care for parents in this situation are often limited (27). This study also recommends that training in how to communicate about DS and deliver a diagnosis must become embedded in the education of all health professionals working in midwifery, paediatrics and neonatology. Ahmed et al. highlights also how midwives in the UK were concerned about the constraints imposed by first trimester combined screening in terms of the limited time in which they had to facilitate informed choice, and the women had to decide about screening. It is crucial therefore to understand healthcare professionals' perceptions of how they communicate the diagnosis of DS to mothers (28).

Türkçapar et al stated recently that for trisomy 21, 89% of obstetricians in Turkey did not define this condition as lethal, and 95% did not consider it incompatible with life. Nevertheless, more than half (57%) always recommended termination (29). Multivariate analysis showed older age and religious beliefs predicted lower termination recommendations for T21; only religious beliefs predicted this for T18. Female obstetricians more often provided palliative care and fetal monitoring for T21.

Kershner et al, in a study published in 1992, through surveys sent by mail to medical professionals in the USA, stated that 63% percent of physicians and 86% of geneticists reported having a biased attitude when informing mothers about the fact that the fetus has trisomy 21 (30). Only 4% of physicians emphasize in the consultation the positive aspects of continuing with the pregnancy of a fetus with trisomy 21 (31). Healthcare professionals must provide accurate, complete, comprehensive, and objective information so that

patients and families can make free and informed decisions about treatment. Following a prenatal diagnosis, counselling for parents should include reliable estimates of survival and quality of life (32). The knowledge and attitudes of obstetricians influence the quality of the information shared and the parents' decision-making. When a congenital problem is detected and the decision is made to continue the pregnancy, ethical conflicts may arise in the decision-making process regarding pregnancy and childbirth (29).

Therefore, we know that the feedback provided by obstetrics teams will have a decisive impact on the parents' decision-making, and on this topic, we have little available literature.

This survey focuses on the quality, balance, and context of the information provided to pregnant women and their families, including whether they receive accurate data on prognosis and quality of life, access to support networks, and sufficient time for reflection. Furthermore, the study promotes communication practices that combine medical accuracy with empathy and respect, recognizing that a physician's words can significantly influence the experience and parental attachment. The aim of this research is not to judge obstetric decisions, but to investigate the prenatal care process. The training of professionals and counselling practices can influence parents' perceptions and choices after a high-risk or confirmed diagnosis of trisomy 21. Pregnancy termination rates following such diagnoses may indicate structural ableism and social pressures that risk devaluing the lives of people with disabilities, potentially leading to indirect discrimination. From the principles of justice and non-discrimination, this study's relevance is significantly enhanced when interpreted through the lens of the UN "Convention on the Rights of Persons with Disabilities" (2006), which explicitly condemns practices that devalue lives based on disability.

In summary, communication of prenatal DS diagnosis in the healthcare setting reveals gaps that can influence pregnant women's decision-making regarding their pregnancy.

The primary objective of our study is to describe the perceptions of obstetricians and gynaecologists on how they communicate the diagnosis of Down syndrome to mothers and what resources they have available to provide this information.

Secondary objectives are:

1) Describe the use of diagnostic tests for chromosomal abnormalities during pregnancy.
2) Describe the prenatal management of information provided to pregnant women in cases of high risk or confirmed diagnosis of Down syndrome.
3) Describe the resources offered to pregnant women in cases of high risk or confirmed diagnosis of Down syndrome.
4) Analyse the differences in the care process between countries, based on the sociodemographic index (SDI).

All procedures will comply with internationally accepted standards for research ethics and authorship. Survey participation is voluntary and anonymous.

Material and Methods

This study is a global, prospective, cross-sectional survey. The study population will consist of healthcare personnel potentially exposed to report a prenatal diagnosis of Down syndrome (obstetricians / gynaecologists / maternal-fetal medicine physicians / midwives).

A survey of 49 questions has been developed using a modified Delphi method (33,34). An online and anonymous reactive Delphi technique was used, where six of the authors (GC, BS, JLG, TP, LM, TV) experts on the topic of the study, from different areas, commented on a previously prepared survey (FG, LC, PG). Consensus on the final version of the survey will be reached after three rounds in which experts will provide modifications to proposed questions and possible answers.

Access to the questionnaire will be provided in English through the Survey Monkey platform (https://www.surveymonkey.com). The questionnaire will be designed to take approximately 15 minutes to complete.

The Ethics Committee of the University of Navarra (Spain) will review the study. This potential approval will cover both the information sheet for the participants and the questionnaire. Respondents will be informed through a cover letter that participation in the survey is voluntary.

The survey includes several types of questions with their respective response options: dichotomous and multiple-choice. Depending on the purpose of the question, some questions allowed only one choice, while others may have one or more alternatives.

The questionnaire includes questions regarding demographic aspects of the obstetrics centre and the responding professional. It then asks about the resources available for prenatal diagnosis at each centre and how this prenatal information is provided in situations with a high suspicion of trisomy 21 in the fetus. Finally, it poses questions about the obstetrician's self-perception of how they provide this information, as well as the resources available to families regarding Down syndrome.

The survey will be distributed through obstetric societies, working groups, authorized email lists, general and obstetric social and professional networks (http://www.linkedin.com). To facilitate successful recruitment, we will utilize professional social networks. We will also contact medical centers and specialized clinics, seeking direct contact with specialists to foster trust in the research process (35). Our research group has previous experience in the global and continental distribution of surveys in the field of neonatology, with a good capacity for dissemination (36–41). The questionnaire is available at the end of this document. The survey will be available from January to June 2026.

The survey data will be grouped for analysis according to each country's SDI (11). SDI is calculated using the geometric mean of three key indicators: per capita income, the

average number of years of education for individuals aged 15 and older, and total fertility rate, which represents the average number of children a woman is expected to have in her lifetime. SDI values are scaled to a range of 0 to 1, with 0 equalling the lowest income, lowest schooling, and highest fertility rate, and 1 equalling the highest income, highest schooling, and lowest fertility rate assessed during that time (42,43). For comparisons across SDI quintiles (low, low-middle, middle-high, and high SDI), each country will be assigned to a single quintile according to its SDI in 2019. Because there may be few responses from countries with low SDI, it is possible to combine both low and low-middle SDI countries into a single category for analysis.

The frequencies and percentages of categorical variables will be calculated for analysis. The statistical relationship between SDI and other variables is related. On the other hand, the Chi-square test is applied to examine relationships between categorical variables. To control family-wise Type I error, we will set the significance level for the analyses at $p<0.05$. Statistical analysis will be conducted using IBM SPSS Statistics®, version 29.

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Participants information sheet (to be added at the beginning of the survey)

First, we thank you for your interest in participating in this study.

We invite you to participate in an investigation on the perceptions of obstetricians and gynaecologists on how they communicate the diagnosis of Down syndrome to mothers and what resources they have available to provide this information. Secondly, we want to analyse the differences in the care process between countries, based on the sociodemographic index (SDI). The study has been approved by the Research Ethics Committee of the University of Navarra (Spain). Before you decide if you want to participate in this study, it is important that you understand why this research is interesting, what your participation will involve, how your information will be used.

Please take the time to carefully read the information provided below.

What/Why is the reason for the study? we want to know more about the antenatal management of pregnancies with high risk or confirmed trisomy 21. We know that the feedback provided by obstetrics teams will have a decisive impact on the parents' decision-making, and on this topic, we have little available literature.

How will the study be done? Using a questionnaire specifically designed by the researchers and validated following the Delphi methodology.

You should know that your participation in this study is voluntary. Once you answer our questionnaire, since your answer is not identifiable, it will not be possible to delete it.

Who can participate? This aims to be GLOBAL research, and all obstetrician related to the care of pregnancies are welcome to answer the questionnaire. Do not hesitate to share this survey with your colleagues.

Your participation in the study is limited to answering the questionnaire. We do not collect the IP (internet protocol address) from which you reply.

No type of financial compensation is foreseen during the study.

Financing: This study does not have any external financing.

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1. Before we start, do you normally care for mothers during pregnancy as an obstetrician / gynaecologist?
    a. Yes
    b. No
2. Are you?
    a. Female
    b. Male
    c. I prefer not to answer
3. Your age is…Add a number, please
    a. Number
4. Are you a training doctor in obstetrics or gynaecology?
    a. Yes
    b. No
5. For how many years have you been working as a specialist?
    a. Less than 10 years
    b. Between 10 and 20 years
    c. More than 20 years
6. In which country is your hospital located?
    a. List of countries
7. How would you classify the level of care of your obstetric unit?
    a. Level 1. Basic care.
    b. Level 2. Specialty care.
    c. Level 3. Subspecialty care.
    d. Level 4. Regional perinatal health.
8. How would you describe your main current professional activity?
    a. I work for a public hospital / medical centre
    b. I work for a private hospital / medical centre
    c. I work in both public and private hospitals
9. Do you have any teaching activities associated with your obstetric clinical practice? (More than one answer is possible).
    a. I teach in the Medicine degree program
    b. I teach in the Nursing degree program
    c. I teach in the Midwifery degree program
    d. I teach in the Genetics degree program
    e. I do not have any teaching responsibility
10. In your daily clinical practice, do you have the responsibility of training doctors in obstetrics/gynaecology?
    a. Yes
    b. No
11. Do you have any relatives or close friends with Down syndrome?
    a. Yes
    b. No
12. Do you have any relatives or close friends with a congenital malformation since childhood?

a. Yes
b. No

13. Before suggesting screening tests, do you check if the patient knows what Down syndrome is?
    a. Yes
    b. No
    c. Sometimes
14. Which of the following first-trimester screening tests are you most likely to offer?
    a. Maternal serum screening (free β-hCG and PAPP-A)
    b. NT ultrasonography
    c. Combined screening (NT and maternal serum screening)
    d. I do not offer these screening tests
    e. I cannot offer these screening tests in my clinical environment
15. Which of the following second-trimester screening tests are you most likely to offer?
    a. Quad screen (MSAFP, hCG, unconjugated estriol, inhibin-A)
    b. Triple screen (MSAFP, hCG, unconjugated estriol)
    c. Double screen (MSAFP and hCG)
    d. Integrated first- and second-trimester serum screening
    e. I do not offer these screening tests
    f. I cannot offer these screening tests in my clinical environment
16. You routinely offer amniocentesis…:
    a. In all pregnancies at elevated risk for any genetic abnormality, including advanced maternal age
    b. To all patients of advanced maternal age
    c. To all patients
    d. Do not offer the procedure routinely
17. You personally perform amniocentesis…:
    a. On singleton pregnancies
    b. On singleton and multiple pregnancies
18. How many amniocenteses did you perform in the past year?
    a. Number
19. You routinely offer chorionic villus sampling…:
    a. In all pregnancies at elevated risk for any genetic abnormality, including advanced maternal age
    b. To all patients of advanced maternal age
    c. To all patients
    d. Do not offer the procedure routinely
20. You personally perform chorionic villus sampling …:
    a. On singleton pregnancies
    b. On singleton and multiple pregnancies
21. How many chorionic villus sampling did you perform in the past year?
    a. Number

22. If the combined first-trimester screening test shows an intermediate or high risk of trisomy 21, do you explain what the syndrome is?
    a. Yes
    b. No
    c. Sometimes
23. When a mother pursues a prenatal screen or diagnostic test for Down syndrome, which of the following best describes your typical practice:
    a. At the time of ordering the test, we set up a future time to discuss the result.
    b. When the results come back, I then call her by phone with the results.
    c. When the results come back, I send her the results electronically through our hospital's patient messaging system.
    d. When the results come back, I ask my staff to call her to set up an in-person meeting to discuss the results.
24. When you disclose the results of the prenatal testing, which of the following best describes your typical practices?
    a. I disclose the results to the mother alone.
    b. I disclose the results to the mother and father together.
    c. I disclose the results to whomever the mother invites to join the discussion.
25. When you inform a mother that you are certain that the foetus has or may have trisomy 21, which of the following best describes your typical practice)?
    a. After saying hello, I first acknowledge that I am sorry to deliver some bad/difficult news.
    b. After saying hello, I just state the factual results of the screen or diagnostic test.
    c. After saying hello, I state that I have "some unfortunate news."
    d. Other _____
26. After receiving a high risk or positive confirmation test for trisomy 21, which of the following best describes your typical practice?
    a. I always provide the contact information of a local Down syndrome advocacy organization.
    b. I offer the contact information of a local Down syndrome advocacy organization, if the mother is interested.
    c. I do not have any parents' association to which I can refer
    d. I do not usually offer the contact information of a local Down syndrome advocacy organization until after a parent decides to continue the pregnancy.
27. Do you consider that when you offer amniocentesis your pregnant patients, at high risk of having a fetus with trisomy 21, that the baby may in fact not have the trisomy?
    a. Yes
    b. No
    c. Unsure
28. After informing the mother about the high risk or confirmation of the diagnosis that the fetus has trisomy 21, how often do you think the mother comes with a clear idea

of termination of pregnancy and does not want to hear information about Down syndrome?

a. Never
b. Unlikely
c. Sometimes
d. Often
e. Always

29. After receiving a high-risk diagnosis or confirmation of trisomy 21, how frequently do you generally observe that the mother is supported by her family/partner?
    a. Never
    b. Unlikely
    c. Sometimes
    d. Often
    e. Always
30. Is voluntary termination of pregnancy when a fetus has a trisomy 21 an option in the country where you work as an obstetrician?
    a. Yes
    b. No
31. After you detect that your patient's fetus has Down syndrome, which of the following most often described your practice?
    a. I urge the parents to continue the pregnancy.
    b. I emphasize the positive aspects of Down syndrome so that the parents will favour carrying the pregnancy to term.
    c. I try to be as unbiased as possible with their pregnancy options.
    d. I emphases the negative aspects of Down syndrome so that the parents will favour termination of the pregnancy.
    e. I urge parents to terminate the pregnancy.
32. How do you think that quality of life is for a child or adult with Down syndrome and his/her family is (regardless of associated malformations)?
    a. Very poor
    b. Poor
    c. Neutral
    d. Good
    e. Very good
33. Do you think that a child or adult with Down Syndrome can be happy with his/her life?
    a. Definitely
    b. Probably
    c. Maybe
    d. Likely not
    e. No
    f. I do not know

34. Do you feel pressured by your healthcare environment to recommend voluntary termination of pregnancy with a fetus affected by trisomy 21?
    a. Yes
    b. No
    c. Sometimes
35. Do you feel pressured by your political/social environment to recommend voluntary termination of pregnancy with a fetus affected by trisomy 21?
    a. Yes
    b. No
    c. Sometimes
36. Do you offer psychological support services to your pregnant mothers with high risk or confirmed trisomy 21 who wish to terminate the pregnancy?
    a. Yes
    b. No
    c. I don´t have that service available in my centre
37. Do you consider what the quality of the emotional support services offered to your pregnant mothers with high risk or confirmed trisomy 21 is?
    a. Very high
    b. High
    c. Neutral
    d. Poor
    e. Very poor
38. Do you feel that terminating a pregnancy because of a diagnosis of Down syndrome constitutes discrimination against people with disabilities?
    a. Yes
    b. No
39. If you practice a religion, do you believe that it influences in any way your healthcare management of an antenatal high-risk or confirmed case of trisomy 21?
    a. Yes, I believe that my religion/spirituality has an impact on my management
    b. I am a religious/spiritual person, but this has no impact on my management
    c. I am not a religious/spiritual person
40. How would you rate your residency training regarding prenatal screening and diagnosis of fetal aneuploidy?
    a. Non-existent
    b. Inadequate
    c. Barely adequate
    d. Adequate
    e. Comprehensive
41. How qualified do you feel yourself to manage patients in the area of general prenatal genetic counselling?
    a. Not qualified
    b. Somewhat qualified
    c. Well qualified

42. How qualified do you feel yourself to manage patients at elevated risk of aneuploidy counselling?
    a. Not qualified
    b. Somewhat qualified
    c. Well qualified
43. How qualified do you feel yourself to manage patients with a positive screening for fetal aneuploidy counselling?
    a. Not qualified
    b. Somewhat qualified
    c. Well qualified