Information prior to prenatal diagnosis

Knowledge, informational needs and decision-making

ELLEN TERNBY
The overall aim of this thesis was to explore different aspects of information relevant to decision-making regarding prenatal diagnosis (PND) for chromosomal anomalies (CA).

In Papers I–II, women and partners undergoing combined ultrasound and biochemistry (CUB) tests, invasive tests or declining PND for CAs answered a questionnaire. Overall, expectant parents had varying to low levels of knowledge about Down syndrome (DS), with few differences between women and partners, or between those accepting or declining PND. Thus, knowledge at these levels does not seem to influence the decision to accept or decline PND. Some seem to regard CUB as a routine test.

Paper III explored midwives’ perspectives with a questionnaire. The majority believed they had not received sufficient education about PND, and few felt knowledgeable enough to provide information about DS. Most midwives desired more education regarding tests and DS. Actual knowledge levels concerning DS varied, and in some cases, were low.

Paper IV explored the factors influencing decisions concerning PND through interviews with pregnant women. The decision-making process is affected by individual factors (i.e. attitude towards anomalies, worry and need for reassurance, and self-perceived risk) and external factors (i.e. test characteristics and influence from others). The quality of life for an affected individual and the impact on the family is important for some women when making decisions about PND. Healthcare professionals can influence women’s decisions through their attitudes, how they present the tests, and the woman’s individual probability of CAs.

Paper V used Q methodology to explore women’s views on what is important when receiving information about PND. Some women prefer receiving information gradually, while others prefer comprehensive information early in pregnancy. Some value information about the conditions tested for early in the process. The extent to which women wanted to involve their partner in the decision-making process varied. None preferred group information sessions.

In conclusion, providing information and pre-test counselling to pregnant women is a complex task. There is room for improvement in the information provided to expectant parents, and in the education provided to midwives related to PND and DS. Women’s decisions regarding PND are influenced by both individual and external factors. Information about test characteristics and conditions tested for could be helpful for pregnant women when they make decisions. Healthcare professionals’ approach can influence women’s decisions. Women have varying informational needs, making individual and non-directive pre-test counselling with a competent healthcare professional essential to facilitate informed decision-making.

Keywords: Prenatal diagnosis, decision making, chromosome aberrations, genetic counseling, informed consent

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In loving memory of my grandfather Bo, who introduced me to the exciting world of medicine at an early age, and to my family for all your love and support.
List of Papers

This thesis is based on the following papers, which are referred to in the text by their Roman numerals.


IV. Ternby E., Axelsson O., Ingvoldstad Malmgren C., Georgsson S. (2023) Factors influencing pregnant women’s decision to accept or decline prenatal diagnosis - A qualitative study. *Manuscript submitted*


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## Abbreviations

<table>
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<tr>
<th>Abbreviation</th>
<th>Description</th>
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<tr>
<td>PND</td>
<td>Prenatal diagnosis (including both screening and diagnostic tests)</td>
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<td>CUB</td>
<td>Combined ultrasound and biochemistry</td>
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<td>NT</td>
<td>Nuchal translucency</td>
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<td>NIPT</td>
<td>Non-invasive prenatal test</td>
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<td>AC</td>
<td>Amniocentesis</td>
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<td>CVS</td>
<td>Chorionic villus sampling</td>
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<td>CA</td>
<td>Chromosomal anomaly</td>
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<td>DS</td>
<td>Down syndrome</td>
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<td>TOP</td>
<td>Termination of pregnancy</td>
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<td>SBU</td>
<td>The Swedish council on Technology Assessment in Health Care</td>
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<tr>
<td>SMER</td>
<td>Swedish National Council on Medical Ethics</td>
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<tr>
<td>SFOG</td>
<td>Swedish Society of Obstetrics and Gynecology</td>
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Sweden has a tradition of offering comprehensive welfare and healthcare services at little or no cost across its 21 counties. Each year, approximately 105,000–120,000 children are born in Sweden.\textsuperscript{1,2} For most expectant parents, pregnancy is a time of joy and great anticipation, but it can also evoke feelings of worry and concern about the well-being of the fetus.\textsuperscript{3–5} While most children are born healthy, around 2-3\% are born with serious fetal anomalies.\textsuperscript{6,7} Expectant parents may experience a lack of control during pregnancy, and in that context, medical technology and prenatal diagnosis (PND) can be seen as a means to gain better control, reduce uncertainty and gain reassurance about the health of the fetus.\textsuperscript{8–13}

The vast majority of pregnant women have uncomplicated pregnancy outcomes without much intervention. As a result of accessible antenatal care, the perinatal mortality rates globally in developed countries have been reported to be around 5-10 per 1,000 births,\textsuperscript{14} while in Sweden, it is closer to 4-5 per 1,000 births.\textsuperscript{1,2} Sweden’s prenatal screening systems resemble those used in many other healthcare systems in European countries. In Sweden, antenatal healthcare is offered to all pregnant women in outpatient units operated by midwives. Almost all pregnant women attend antenatal care and have regular check-ups (on average eight to ten times) with a midwife during pregnancy.\textsuperscript{15–17} The focus of antenatal care is to promote good sexual and reproductive health, prevent disease in both the pregnant woman and fetus through health education and psychosocial support, and to ensure early detection of complications to provide timely interventions if needed.\textsuperscript{14–16,18} Medical doctors typically become involved only if there are any medical complications or concerns during pregnancy.\textsuperscript{15} Almost all women choose to undergo the second-trimester fetal ultrasound scan, even if they decline other prenatal tests, e.g. combined ultrasound and biochemistry (CUB) and invasive tests.\textsuperscript{6,17,19,20}

Over the past few decades, advancements in medical technology have made PND available not only to high-risk pregnancies but to most pregnant women. Offering and providing information and counselling about available prenatal tests, as well as offering PND, has become standard practice in routine antenatal care. In Sweden, the offer of PND is optional in addition to the standard antenatal healthcare that is routinely provided to all pregnant women.\textsuperscript{15,21}
Background

Information and counselling prior to prenatal tests

Guidelines and regulations

The Swedish law on genetic integrity\(^{22}\) mandates that all pregnant women in Sweden shall be offered general information about fetal diagnostic testing. Additionally, according to the Council of Europe,\(^{23}\) expectant parents should be provided pre-test counselling with sufficient information to enable them to make an informed decision before screening for congenital anomalies. Moreover, the National Board of Health and Welfare in Sweden has published guidelines\(^{21}\) stating that pregnant women shall be offered general information about PND during their first visit to the outpatient antenatal care clinic. Most outpatient antenatal care units adhere to this guideline. If interested, this information is, in most counties in Sweden, provided by the midwife during the first visit. Thereafter, the woman can choose to be referred to a Fetal Medicine Unit or an ultrasound unit to undergo PND for chromosomal anomalies (CA), as well as first- and/or second-trimester ultrasound scans.

The information regarding PND for CAs focuses on the probability of expecting a child with a trisomy, e.g. Down syndrome (DS), and the risks associated with invasive tests. Information about the conditions screened for, i.e. DS and the consequences for the child and the family is not routinely provided. However, according to the Swedish National Council on Medical Ethics (SMER), the information should include both medical and psychosocial aspects.\(^{24}\) In a more recent report, SMER stressed the importance of sufficient pre-test counselling prior to the offer of non-invasive prenatal testing (NIPT). This information should include information on the conditions screened for, such as the medical consequences for the affected child, as well as what it means to live with a child with a CA.\(^{20}\)

Providing information

Research has shown that midwives or physicians are important sources of information for pregnant women about PND.\(^{10,12,13,25,26}\) Providing information about PND and the conditions tested for is far from easy, as it needs to be well balanced and as neutral as possible to avoid inappropriate influence by healthcare professionals.\(^{21,24,27–29}\) In 2007, a literature review concluded that...
some healthcare professionals may not be adequately prepared to provide information on DS and that they need further education about the consequences of DS for affected individuals. More recent studies have indicated that healthcare professionals working in antenatal care and providing information to pregnant women about PND often have important key deficiencies in knowledge related to DS testing. These professionals themselves feel they have inadequate knowledge and request or value more education on the subject. It has been suggested that the nursing and midwifery curricula in European countries needs to be updated, and several reports have stressed that medical and social aspects of the conditions tested for should be included as integral parts of information provided to expectant parents. The importance of facilitating informed choice through rigorous pre- and post-test counselling, as well as ensuring informed consent, regardless of the type of testing performed (invasive or non-invasive), has previously been highlighted. An expert group developed European guidelines for offering prenatal tests, concluding that healthcare professionals must ensure they are well-informed and maintain their knowledge on all relevant aspects of PND.

Prenatal diagnosis for chromosomal anomalies

The medical indication for offering PND is to detect fetal diseases or anomalies, as well as to identify other conditions that may cause complications during pregnancy or affect the preferred mode of delivery (e.g. placenta previa or multiple pregnancy). In many cases, a detected anomaly cannot be treated, and the expectant parents are confronted with the choice to terminate or continue the pregnancy. In other cases, knowledge about a condition before birth provides the opportunity to prepare for appropriate medical care after birth. PND for CAs, such as DS, was introduced in the 1970s. Initially, amniocentesis (AC) in the second trimester was the only method for PND, later followed by chorionic villus sampling (CVS), and more recently by screening-methods such as the CUB test and NIPT. PND for CAs, such as DS, is now frequently used in Sweden as a part of the public antenatal care. The first- and second-trimester fetal ultrasound scans, and CUB tests are usually performed by specially trained midwives, while the invasive tests are performed by a doctor.

Test methods

Ultrasound

The second-trimester fetal ultrasound scan, commonly referred to as the ‘routine ultrasound’, is an examination that is not primarily focused on assessing CAs. It is offered to all pregnant women as part of the antenatal care in
Sweden, and is performed using transabdominal ultrasound at 18-20 gestational weeks. More than 97% of all pregnant women in Sweden choose to undergo this scan.\textsuperscript{19} The reported sensitivity for detection of fetal anomalies with the second-trimester fetal ultrasound and the false-positive rate varies between studies with a sensitivity of 19-80% and a false-positive rate of 0.06-0.5%.\textsuperscript{47} These rates vary depending on the specific condition, as some fetal anomalies are easier to detect than others. In addition to the second-trimester ultrasound, half of the regions in Sweden now offer a first-trimester ultrasound scan\textsuperscript{19} at 11-13 gestational weeks. The purposes of these ultrasound examinations are to determine the gestational age and due date, as well as identify whether it is a singleton or multiple pregnancy, assess the position of the placenta, and to screen for abnormalities of fetal anatomy.\textsuperscript{6,20,44} Ultrasound examinations are considered to have a positive effect on pregnancy surveillance and the planning for delivery.\textsuperscript{6}

Invasive tests
There are two different kinds of invasive tests: AC and CVS. These are the only available diagnostic tests for CAs.\textsuperscript{46} Both methods carry a risk of fetal loss, and have in the past been considered as having a procedure-related miscarriage rate of 0.5-1.0%.\textsuperscript{47,48} However, more recent studies indicate that the risk could be as low as 0.1-0.2%,\textsuperscript{49-51} or even negligible.\textsuperscript{52} The methods for chromosomal analyses are the same for both AC and CVS and usually includes either full karyotyping or the more limited and quicker QF-PCR, which only analyses chromosomes 13, 18, 21, X and Y. Array-CGH, a molecular biological analysis, is increasingly used when there is a need for a more detailed analysis of the chromosomes. It can detect even minor changes in the chromosomes e.g. microdeletions or microduplication syndromes. Depending on the method of analysis used, it takes approximately two days (QF-PCR) or 10-14 days (full karyotyping or array-CGH) to get the test results.\textsuperscript{20,47,53}

Chorionic villus sampling
CVS is the method of choice for obtaining fetal tissue for diagnostic PND before 15 gestational weeks.\textsuperscript{47,54} It can be performed from 11 gestational weeks. During the procedure, a sample is taken from the placenta transabdominally using a needle guided by transabdominal ultrasound.\textsuperscript{47}

Amniocentesis
The amniotic fluid contains cells shed by the fetus and can therefore be used for chromosomal analyses. AC can be performed from 15 gestational weeks.\textsuperscript{47,55} With ultrasound guidance, a sample of amniotic fluid is aspirated transabdominally through a needle from the amniotic sac. Subsequently, a chromosomal analysis is performed on the amniotic fluid.\textsuperscript{47}
**Combined ultrasound and biochemistry test**
The CUB test provides an individual probability assessment of CAs, rather than a definitive diagnosis. For the past 20 years, it has been considered the method of choice for probability assessment since it has had the best balance between the number of detected cases and false-positive results. The CUB test consists of two parts: ultrasound nuchal translucency (NT) measurement and maternal serum biochemistry. In addition, maternal and gestational age are also taken into consideration when assessing the probability of a CA. With an early ultrasound scan at 11-13+6 gestational weeks, an accumulation of fluid at the back of the fetus’s neck (NT), is measured. An increased NT measurement, i.e. an increased amount of fluid, is associated with a higher probability of CAs, such as DS. Maternal biochemical screening is performed through a blood sample from the expectant mother (taken at the earliest at nine gestational weeks) from which free human chorionic gonadotropin (β-hCG) and pregnancy associated plasma protein (PAPP-A) are analysed. These substances are produced in the placenta, and their levels increase and decrease, respectively in cases of DS pregnancies. An increased probability with CUB requires follow-up testing for a definitive diagnosis. The frequency of detected DS pregnancies with CUB depends on the cut-off level for offering diagnostic tests, and is also influenced by the population that is offered the CUB test. Approximately 5% of all CUB tests will result in a CUB probability greater than 1:200. Among these 5%, 85-90% of all DS pregnancies are found.

**Non-Invasive Prenatal testing**
Cell-free fetal DNA originating from the placenta can be detected circulating in the maternal plasma during pregnancy. In recent years, new methods of analysing fetal genetic material from a maternal blood sample have been developed. NIPT for fetal trisomies 13, 18 and 21, as well as analyses of the sex chromosomes, is now available in many countries. In Sweden, the analyses do not always include the sex chromosomes, and some counties only analyse chromosomes 13, 18 and 21. The test has a high specificity and sensitivity for trisomies 13, 18 and 21, but it is not considered a diagnostic test due to variations in positive predictive value (PPV). Therefore, NIPT is recommended only as a high-specific screening method. The results are highly reliable in case of a screen negative result, but in case of a screen positive result, it requires follow-up diagnostic testing. NIPT is now considered the most accurate screening test for trisomies 13, 18 and 21 in unselected singleton pregnancies, as well as pregnancies with a known increased probability. Furthermore, NIPT has several advantages compared to previously available prenatal tests; for example, a maternal blood sample is a simple and risk-free procedure, and it can be performed already from gestational week 10.
How the different tests are offered

In Sweden, the offer and availability of PND varies among the 21 counties. Since there is a significant increase in the probability of trisomies with increasing maternal age, pregnant women aged 35 or older have traditionally been offered PND with AC or CVS despite the procedure-related miscarriage risk. Thus, in the past, the majority of invasive tests were performed due to advanced maternal age (approximately 75%). However, some tests were performed in younger women (<35 years) due to worry and some were motivated by a known increased risk of a hereditary CA, monogenic diseases or by abnormal ultrasound findings.

A systematic review by the Swedish Council on Technology Assessment in Health Care (SBU) in 2006 concluded that conducting a risk assessment with a CUB test prior to invasive testing is preferable compared to relying solely on the woman’s age for assessment. One year later, SMER came to the same conclusion. Despite these recommendations, there has been a lack of consistent national guidelines, and the offer of prenatal tests still vary among the counties of Sweden. In 2011, 14 out of the 21 counties in Sweden offered the CUB test, while one county offered the NT test to pregnant women. Today, all but three counties offer the CUB test, but how the test is offered differs among the counties. Some offer it to all pregnant women, while others have an age limit, and the cost of a test can vary. The number of pregnant women in Sweden undergoing a CUB test has steadily increased over the years. Today, approximately 70% of women undergo screening for CAs with a CUB test.

When a pregnant woman has a high CUB probability, she is offered either NIPT for further screening, or invasive testing to get a definite diagnosis through chromosomal analyses. The cut-off levels for when diagnostic tests have previously been offered have varied among the counties in Sweden, but have usually been set to a probability for DS between 1:200 and 1:400. These cut-off levels are a pragmatic consideration involving several factors. Setting a higher cut-off level means fewer patients will be considered for invasive tests, which will result in the detection of fewer DS pregnancies, and in turn, generate fewer procedure-related miscarriages.

In recent years, the use of NIPT has increased rapidly around the world, and it is now offered in most of the counties in Sweden (19 out of 21). There is an on-going discussion on how NIPT should be offered. The cost of NIPT is still relatively high compared to screening with CUB, which often prevents it from being offered as a first-tier test to everyone. Therefore, it is more often used as a second-tier test after a previous screening with CUB. SBU has suggested that NIPT should be offered to women with a previous CUB test with a probability of a trisomy greater than 1:200. Another strategy suggested by the Swedish Society of Obstetrics and Gynecology (SFOG) is to offer NIPT to women with a CUB probability of 1:51-
1:1000, while offering invasive testing directly to those with a CUB probability greater than 1:50. The majority of Swedish counties now offer NIPT as a second-tier test following a high CUB probability (with varying cut-offs of 1:51-1:200/1:300/1:1000), while two counties offer it as a first-tier screening test to older women without a previous CUB test. As expected, since the introduction of non-invasive screening methods, i.e. CUB and NIPT, the number of invasive tests has decreased in Sweden during the last decade. In 2021, approximately 1.2% of women who gave birth had undergone invasive testing. However, invasive tests still remain the only diagnostic option for CAs.

Chromosomal anomalies

There are numerous types of CAs, with a wide variety of phenotypes. Trisomies 13 (Patau’s syndrome), 18 (Edward’s syndrome) and 21 (Down syndrome (DS)), as well as sex chromosome anomalies involving chromosomes X or Y (Turner’s syndrome (45;X), Klinefelter’s syndrome (47;XXY) and XYY syndrome), together account for the majority of all CAs detected by the routinely offered PND. Patau’s and Edward’s syndromes are very severe conditions associated with various congenital malformations. Children affected by trisomy 13 or 18 rarely survive more than a few days to weeks after birth, and just under 10% survive their first year. DS has a milder manifestation and is thus the most common of the trisomies, as the more severe trisomies often result in spontaneous abortions. The symptoms of sex chromosome anomalies are considerably milder, and many individuals with these anomalies live a normal life without significant physical or intellectual disabilities. Since DS is the most common CA with clinically significant symptoms, it has been the main focus of prenatal screening and diagnosis, and it is also the chromosomal condition that many expectant parents have heard of. The reported incidence rates for the more severe CAs vary between countries, but in Sweden, it is approximately 0.5-1 in 10,000 live births for trisomy 13 and 1-2 in 10,000 live births for trisomy 18. The incidence rates of the sex CAs have been reported to be approximately 1 in 600-700 male live births for Klinefelter’s syndrome, 1 in 2,000-2,500 female live births for Turner’s syndrome, and 1 in 1,000 male live births for the XYY syndrome.

The probability of expecting a child with a trisomy, such as DS, increases with advancing maternal age. This probability rises after 30 years of age and more significantly after 40 years. The average age of women giving birth has increased over the past decades, both in Sweden (where one out of four pregnant women are aged 35 years or older), as well as in most of the western world. Despite this trend, until recently, the number of DS births in Sweden has remained at the same level since the late 1990s, i.e. approximately
1 in 700-800 live births.\textsuperscript{7,88–90} This stability may be explained by the increasing use of PND, the subsequent increase in prenatally diagnosed CAs and an increasing number of termination of pregnancies (TOPs) during the same period.\textsuperscript{7,64,88,89} While reports from other parts of the world have indicated a similar trend, some countries have previously experienced a decrease in the incidence of DS births.\textsuperscript{45,88} In 2015-2016 a decrease in the number of DS births was observed in Sweden as well, with an incidence closer to 1 per 1,000 live births.\textsuperscript{7} Approximately 60% of all children born with DS in Sweden between 1999 and 2015 had mothers that were younger than 35 years old, an age group that, at the time, was not routinely offered PND for CAs in many Swedish counties.\textsuperscript{7,64} In 2016, the corresponding number was 50%, which may be a reflection of more women under 35 years being offered prenatal screening with CUB, resulting in more DS pregnancies being diagnosed and terminated.\textsuperscript{7} For trisomies 13 and 18, rates for TOP are high, in part due to higher diagnosis rates and the severity of the conditions.\textsuperscript{7}

**Down syndrome**

DS is the most common cause of intellectual disability. There are four kinds of CAs resulting in DS,\textsuperscript{91–93} which is always caused by excessive chromosomal material from chromosome 21. The most common type, accounting for approximately 94% of all DS cases, is the result of a failure of chromosomes to separate properly during meiosis (non-disjunction), resulting in an extra chromosome 21 in all cells, i.e. a trisomy.\textsuperscript{91,92,94,95} Little is still known about the cause of the non-disjunction.\textsuperscript{45,92} Apart from maternal age, there are no known environmental factors contributing to the occurrence of DS, and it is found in all countries and cultures.\textsuperscript{88,91,92,96} The incidence, however, varies between countries from 1 in 500 to 1 in 1,000 live births.\textsuperscript{91} This variation in incidence is mostly due to differences in maternal age,\textsuperscript{91} but probably also because of the availability of PND, and TOP, as well as available support for individuals with disabilities. Previously, approximately 120-140 children with DS were born in Sweden each year, which corresponds to 1 child with DS per 700-800 live births.\textsuperscript{7,88–90} As mentioned, according to the latest reports, this has decreased to approximately 1 child per 1,000 live births.\textsuperscript{7} The syndrome is associated with some level of intellectual disability and developmental challenges, and there are often medical and social consequences.\textsuperscript{92} There is a large variation in both the level of intellectual disability and the extent of medical complications. In the past decades, there has been a progress in the treatment of the medical issues in individuals with DS, and the mean survival age has increased to around 60 years.\textsuperscript{90,89,91,97} Practically everyone with DS learns to walk.\textsuperscript{98} Medical comorbidities include congenital heart defects (most commonly atrioventricular septal defects, ventricular septal defects or atrial septal defects), which are seen in approximately 50% of individuals with DS (compared to a baseline risk of around 1%) and often require
surgical rectification.\textsuperscript{90,99–102} Gastrointestinal anomalies (mostly duodenal atresia) that require surgical rectification in the newborn period are found in about 10%.\textsuperscript{90,102} Hearing problems and chronic ear infections are common. The reported frequency of hearing problems has varied, but studies suggest that if chronic ear infections are left untreated, as many as 70% will develop hearing problems.\textsuperscript{97,102,103}

There has also been a significant development in the pedagogical treatment of children with DS. Delays in language skills are not uncommon,\textsuperscript{104} which may be partly due to hearing problems and affected oral-motor structure and function, but also a reflection of more general delays in cognitive development.\textsuperscript{105} Research has indicated that the verbal and academic skills are influenced by environmental factors, and whereas only a few individuals with DS learned to read in the past, these skills have improved nowadays.\textsuperscript{91,98,105}

Families with a child with DS do not notably differ from other families regarding social aspects.\textsuperscript{103} More than 90% of children with DS live with their biological parents,\textsuperscript{89,91} and they have siblings to the same extent as in other families.\textsuperscript{103,106} The frequency of divorces does not seem to differ between parents with and without DS children.\textsuperscript{98,103,106–108} Swedish studies have not shown differences in the total employment rate when comparing parents of children with DS to other parents, but mothers of children with DS more often work part-time.\textsuperscript{107,109} In the past, most people with DS lived in institutions, but today, a majority of grown-up individuals are able to live on their own with individually tailored support.\textsuperscript{91,95}

Informed choice

According to the Swedish Health and Medical Services Act, good medical care should be based on respect for the patient’s autonomy and integrity, and as far as possible, be performed in accordance with the patient’s wishes.\textsuperscript{110} Thus, to protect the patient’s autonomy and to offer PND in a way that is ethically justifiable, it is important to facilitate informed choices for pregnant women. This is emphasised by a report from the World Health Organisation (WHO), stating that healthcare during pregnancy should be offered based on the woman’s needs, after discussions with healthcare professionals to enable informed choices.\textsuperscript{111} Partners are influential persons when decisions are to be made about the pregnancy, and in Sweden today, most women bring their partners when they receive information on PND. Although the pregnant woman ultimately makes the decision about PND, partners are often involved in the decision.\textsuperscript{11,13,25,28,31,47,112} Consequently, healthcare professionals may have to take the couple’s autonomy (and not just the woman’s individual autonomy) into account when providing information and counselling to expectant parents.
Informed choice is commonly defined as a decision that is based on relevant information and knowledge, is consistent with the decision-maker’s values and behaviourally implemented, i.e. an informed choice should be value-consistent and based on sufficient knowledge.\textsuperscript{113} Informed decision-making in the context of prenatal screening and diagnosis can, however, be challenging, as expectant parents may be forced to make several difficult decisions within a restricted time-period.\textsuperscript{114} This decision-making chain often begins with the choice to accept or decline prenatal screening with CUB or NIPT, a decision that in case of a high probability result is followed by a decision on invasive testing, and eventually the final decision regarding TOP or not if a CA is diagnosed.

Many studies and discussions on informed choice in the context of PND have primarily focused on information and knowledge about the testing method and probability-assessment, with limited focus on information and knowledge regarding the specific condition being screened for. There are indeed issues concerning insufficient information and knowledge about the tests among pregnant women.\textsuperscript{12,115–120} However, information about the condition tested for is also important in the context of informed choice.\textsuperscript{31,40,41,121–123} A few studies have indicated that pregnant women often feel that they have insufficient knowledge about DS,\textsuperscript{31,39,124} and many pregnant women actually have insufficient knowledge about DS and its consequences.\textsuperscript{27,117,125–129} Furthermore, women undergoing PND may not always know what conditions the test can detect.\textsuperscript{31,116} Thus, women’s knowledge is not always sufficient to make informed decisions.\textsuperscript{47,117,120,130–135} Moreover, women do not always make value consistent choices, which can also hamper informed choice.\textsuperscript{117,131,134–136} Research suggest that women with higher levels of education more often make informed decisions.\textsuperscript{134}

Studies have indicated that pregnant women who make informed choices concerning PND have improved psychosocial outcomes and experience less decisional conflict.\textsuperscript{121,131,134,136–140} Concerns have been raised that participation in screening might increase anxiety, however, in the literature, there is no evidence of increased anxiety in women receiving a screen-negative result. This indicates that participation itself does not lead to persistent increased anxiety, although a screen-positive result might.\textsuperscript{4,133,139,141} In the context of prenatal testing, information and increased knowledge do not necessarily lead to increased worry among pregnant women,\textsuperscript{47,133,142} but rather to higher levels of wellbeing and less anxiety, decisional conflict and regret concerning the decision.\textsuperscript{138,143} Women who are not satisfied with the genetic counselling they have received are more likely to experience decisional conflict and subsequent decisional regret.\textsuperscript{144}
Women’s informational needs

When receiving information prior to decisions about PND, pregnant women value information regarding risks associated with the different tests. Besides information about test characteristics, they also desire information about DS and the probability of expecting a fetus with DS early in pregnancy, prior to PND. Women’s informational needs may be affected by their probability of expecting a fetus with a CA in a previous or current pregnancy. Furthermore, women who have already made their decision to undergo or to abstain from PND when attending pre-test counselling may not need as much information as those who are still undecided. There is a need for further research investigating expectant parents’ informational needs and preferences, both regarding content and ways of receiving it.

A pregnant woman undergoing PND may find herself in a situation where she must deal with distressing information and make difficult decisions. Two psychological coping styles for dealing with potentially threatening medical situations, such as PND, have previously been described: ‘monitoring’ i.e. focusing on and seeking threat-relevant information and ‘blunting’ i.e. avoiding information relevant to the threat and seeking distraction. In various medical fields, studies have found an association between coping styles and informational needs, and satisfaction with information.

Decision-making regarding prenatal diagnosis

The decision-making process for pregnant women regarding PND is influenced by different aspects. A recent review of quantitative research described several levels of influence, with an individual level affected by demographic, clinical and psychological aspects; a relational level with influence from family (especially partners) and society; and a contextual level influenced by information received and test characteristics. According to this review, one of the main reasons for having PND is advanced maternal age. A previous review, which also included qualitative studies, reported similar findings, where the decision-making process was influenced both by test characteristics and by individual factors such as age, anxiety, personal or professional experience of and attitude towards disability and TOP. Many reports indicate that worry and need for reassurance of a healthy baby can motivate having PND. Additionally, many studies have also highlighted the influence of partners on decisions regarding PND. Qualities of the test, in particular test-related risks, but also test accuracy, easiness and timing seem to be important considerations for women when they make decisions about PND.

In addition, the perceived quality of life for a child with an anomaly, as well as its impact on parents’ and family life, seems to be important factors.
affecting decision-making regarding PND.\textsuperscript{40,41,145,153,155,156} Parental responsibilities have been described by pregnant women (both women accepting and declining prenatal screening) as context dependent, acknowledging that their decision could have been different under other circumstances.\textsuperscript{156} Hence, it is not surprising that the decision-making process following a diagnosis of DS has been reported to be influenced by various demographic factors (such as maternal age, gestational age, number of already existing children, history of induced abortion and religion) as well as by psychosocial factors (such as perceived parenting burden/reward, quality of life for a child with DS, attitudes towards individuals with disabilities, and support from others).\textsuperscript{157}
Aims

The overall aim of this thesis was to explore different aspects of information and knowledge about DS linked to PND for CAs.

The specific aims of each study were:

I To assess why pregnant women and partners decided to undergo a CUB test, the information they have received and how they perceived it, as well as assess their knowledge about DS, and their thoughts about invasive procedures and the possibility of TOP.

II To investigate whether knowledge of DS influences pregnant women’s decision to accept or decline PND for CAs. Further aims were to explore reasons for accepting or declining PND and to discern differences between those who accept and those who decline PND concerning the information received, knowing someone with DS and thoughts about their decision-making.

III To investigate what information midwives provide to expectant parents prior to PND, expectant parents’ requests for information, midwives’ knowledge of PND and detectable conditions, i.e. DS, and midwives’ perceptions of their own competence in providing information.

IV To investigate what factors influence pregnant women’s decisions to accept or decline PND. Further, to explore their views and reflections on PND and detectable conditions, e.g. DS.

V To study pregnant women’s subjective viewpoints regarding what is important when receiving information prior to decision-making about PND for CAs. A secondary aim was to investigate whether informational needs differ according to different socio-demographic backgrounds and coping styles.
## Materials and methods

### Overview of included Papers (I-V)

Table 1. An overview of the five included papers regarding aims, design, participants and methods of data analysis.

<table>
<thead>
<tr>
<th></th>
<th>Aim</th>
<th>Design</th>
<th>Participants</th>
<th>Data analysis</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Assessing expectant parents’ knowledge of DS and information received prior to CUB testing and their reason for undergoing CUB.</td>
<td>Quantitative, cross-sectional study based on a questionnaire.</td>
<td>105 pregnant women and 104 partners attending a CUB test in Uppsala.</td>
<td>Descriptive and comparative statistical analyses.</td>
</tr>
<tr>
<td>II</td>
<td>Investigating reasons for accepting or declining PND and the effect of knowledge of DS. In addition, to compare information, experience of DS and thoughts about decision-making between acceptors and decliners.</td>
<td>Quantitative, cross-sectional study based on two questionnaires.</td>
<td>76 pregnant women undergoing invasive tests and 65 women declining PND for CAs in Uppsala.</td>
<td>Descriptive and comparative statistical analyses.</td>
</tr>
<tr>
<td>III</td>
<td>Exploring midwives’ knowledge of DS and prenatal tests and what information they provide to expectant parents.</td>
<td>Quantitative, cross-sectional, study based on a questionnaire.</td>
<td>64 midwives working in outpatient antenatal care in Uppsala.</td>
<td>Descriptive and comparative statistical analyses.</td>
</tr>
<tr>
<td>IV</td>
<td>Exploring what factors influence pregnant women’s decision-making regarding PND.</td>
<td>Qualitative study based on semi-structured interviews.</td>
<td>24 pregnant women in the first trimester (prior to making decisions about PND) in Eskilstuna.</td>
<td>Qualitative content analyses.</td>
</tr>
<tr>
<td>V</td>
<td>Investigating pregnant women’s views on what is important when receiving information regarding PND. Additionally, to investigate whether informational needs differ with different socio-demographic backgrounds and coping styles.</td>
<td>A Q methodological study combined with a set of 2 questionnaires.</td>
<td>45 women in early pregnancy, booking their first appointment at an antenatal care unit in Uppsala, Eskilstuna or Örebro.</td>
<td>Q factor analysis and comparative statistical analyses.</td>
</tr>
</tbody>
</table>
Study setting – Recruitment from three regions

As previously mentioned, the offer of PND varies across different regions in Sweden. The studies included in this thesis have recruited participants from three regions: Uppsala, Eskilstuna and Örebro, each with varying routines for offering PND, as described below. Participants for Papers I-III were recruited in Uppsala. Paper IV recruited pregnant women from Eskilstuna. Participants for Paper V were recruited from all three regions.

**Uppsala (Papers I-III and V)**

At the time of data collection for Papers I-III, Uppsala County had a catchment area with about 323,000 inhabitants, encompassing both urban and suburban areas. By the time of Paper V, this population had increased to around 400,000 inhabitants. The region’s outpatient antenatal care comprised 19 units during the recruitment for Papers I-II, and gradually increased to today’s 28 units. There are around 4,000-4,300 deliveries annually at Uppsala University Hospital.²

During the data collection for Papers I-III, the CUB test was offered to women over 35 at a subsidised cost of 30 € (corresponding to the fee for a doctor’s appointment), while younger women paid a higher cost of 160 €. Women >35 years could choose to have an invasive test without a prior CUB test. Younger women were eligible for invasive tests, when possible, based on the capacity of the Fetal Medicine Unit. In addition to a high CUB probability, other reasons such as advanced maternal age, a previous pregnancy with a CA, ultrasound findings and anxiety could motivate an invasive test. The cost of an invasive test was 30 € regardless of age. At the time of inclusion of participants for Papers I and II, the acceptance rate of CUB in Uppsala County was 30%, consistent with the national figure (26% of expected pregnancies) reported for 2011 in Sweden.⁶⁶

During the recruitment of participants for Paper V, all pregnant women were offered CUB at a subsidised cost (approximately 26 €), regardless of their age. At this time, NIPT was offered as a second-tier test to women with a CUB probability of 1:50-1:200. First-trimester ultrasound scan was not offered.

**Eskilstuna (Papers IV-V)**

Eskilstuna, situated in Södermanland County, has a catchment area comprising approximately 140,000 inhabitants, encompassing both urban and suburban areas. The outpatient antenatal care in Eskilstuna consists of one main unit, where the midwives, in addition, occasionally work at five primary healthcare centres within the area. At Eskilstuna hospital, there are around 2,000-2,200 deliveries annually.²⁷⁹ During the study period for Papers IV and V, CUB and invasive tests were available to pregnant women >35 years, free of charge. First-trimester ultrasound was not offered during the recruitment
period for participants in Paper IV. Since then, however, Eskilstuna started to offer first-trimester ultrasound, which was available to all pregnant women free of charge during the recruitment period for Paper V. NIPT was offered to certain women based on an assessment from a doctor.

Örebro (Paper V)
Örebro County has a catchment area with just above 300,000 inhabitants, incorporating both urban and suburban areas. The region’s outpatient antenatal care consists of 25 units, and there are around 3,000-3,400 deliveries annually at Örebro University hospital. Örebro has developed a special model for informing and offering prenatal screening and diagnosis (the Freja model). Pregnant women are asked by their regular midwife at the outpatient antenatal care unit at their first appointment early in pregnancy, if they are interested in receiving information about PND. Women who wish to receive information are scheduled for a separate appointment with a specially trained midwife (a ‘Freja midwife’) to receive information about PND before booking an appointment at the ultrasound unit, if desired. All pregnant women in Örebro County are offered a choice between a first-trimester ultrasound or a CUB test free of charge. NIPT is offered as a second-tier test at no cost to women with a CUB probability higher than 1:1000.

Procedure and participants
Papers I and II
Three similar questionnaires (one for expectant parents undergoing a CUB test, one for pregnant women undergoing invasive tests, and one for pregnant women declining PND for CAs) were developed specifically for these two studies. These questionnaires were based on current literature and the combined expertise within the research team. Each questionnaire had three sections, with a total of 38-41 questions. The first section of all three questionnaires was the same, containing socio-demographic questions, e.g. age, marital status, previous children and education. The focus of the second section was information provision and decision-making. The questions concerning received information and satisfaction with that information were the same for all three groups, while the questions on decision-making varied. The questions regarding decision-making explored reasons for opting for a CUB test, invasive tests, or declining tests for CAs. For the expectant parents undergoing CUB, there were additional questions regarding decision-making in the event of an increased probability of DS, or a DS diagnosis. The same questions on decision-making in the event of a DS diagnosis were included for the women undergoing invasive tests. The third section was the same for all three groups and included knowledge questions regarding DS and its medical, cognitive
and social consequences. Most questions were of multiple-choice character, but there was room for written comments.

In Paper I, both pregnant women and partners were invited to participate while attending the Fetal Medicine Unit at Uppsala University Hospital for a CUB test between November 2010 and March 2011. Most participants were couples, where both the pregnant woman and the partner completed the questionnaire independently. The questionnaires were anonymous, without any connection between the couples. As some women were included without their partners, some partners were later invited to participate separately to attain an equal number of women and partners. Couples were instructed to complete the questions independently without discussing them.

In Paper II, pregnant women who attended the Fetal Medicine Unit at Uppsala University Hospital to undergo invasive tests, and women who had abstained from PND for CAs, between November 2010 and April 2011, were invited to participate. Women in the invasive test group were included when they had the invasive test at 13–17 weeks of gestation, while those who declined PND were included when they underwent their second-trimester fetal ultrasound scan at 16–18 weeks. To ensure that the women in the declining group had received information and had been offered PND for CAs, and thus had actively made a decision to decline it, all participants included in this group were older than 35 (with the exception of two women). The majority (83%) of the women in the invasive group had not had a CUB test prior to the invasive test.

For both papers, participants meeting the inclusion criteria (Swedish-speaking and aged >18 years) were invited sequentially, whenever possible, based on available clinical resources. All questionnaires were filled in anonymously, and no record was kept of non-participants. The questionnaires were administered at the Fetal Medicine Unit and filled in directly after receiving the results of CUB or the second-trimester ultrasound, but before receiving the results of invasive tests.

Paper III

Based on the current peer-reviewed literature and the combined expertise held by the research team, a five-page questionnaire was specifically developed for the study to cover various aspects of PND and DS from midwives’ perspectives. It consisted of 4 sections, with a total of 33 questions. The first section included socio-demographic characteristics, such as age, year of graduation and years working as a midwife. The second section focused on the information midwives provided to pregnant women and partners prior to PND, what information expectant parents requested and whether the midwives themselves felt they had sufficient knowledge to provide such information. The third section contained the same knowledge questions concerning cognitive, medical and social consequences of DS, as used in Papers I and II. The
last section focused on the education the midwives had received about DS and the different tests, whether they wanted more education, and their views on the current procedures for providing information and how the routines might be improved. As in Papers I and II, most questions were of multiple-choice character, with space for free comments.

Midwives working in the outpatient antenatal care in Uppsala County were offered the opportunity to participate and complete the study-specific questionnaire during an in-service training event in November 2012. Everyone present accepted to participate, and the questionnaire was completed anonymously by 64 out of 70 midwives (91%) working in the outpatient antenatal care in the county.

Paper IV

Paper IV had a qualitative approach and was based on semi-structured interviews with pregnant women. To ensure that the same topics would be discussed, while still allowing some flexibility during the interviews, a semi-structured interview guide (Appendix 1) was developed. This guide was based upon the current peer-reviewed literature and the collective clinical experience of the research team (an obstetrician, a midwife, a genetic counsellor and the author of this thesis). It covered various aspects of women’s informational needs and decision-making process regarding PND, as well as their perceptions of healthcare professionals’ attitudes during pre-test counselling and their own attitudes towards anomalies and DS. The guide was tested through pilot interviews with four pregnant women, who were included in the study since no significant changes were needed for the guide itself or the study set up.

Pregnant women were invited to participate when they called to schedule their first appointment with a midwife at the outpatient antenatal care in Eskilstuna from October 2016 to March 2017. Midwives invited pregnant women meeting the inclusion criteria (pregnant in the first trimester, aged >18, with a good command of the Swedish language) consecutively until saturation was reached. Interested prospective participants received further oral and written information from the first author over telephone. Twenty-four pregnant women accepted participation and were interviewed over telephone on separate occasions. Interviews lasted 15 to 37 minutes (mean 25 minutes). By the twentieth interview, recurring similarities and patterns were noticed by the interviewer. Saturation was confirmed after another four interviews, after which inclusion of participants was discontinued.

Paper V

For Paper V, Q methodology was used to explore pregnant women’s subjective viewpoints on what is important when receiving information regarding PND, concerning both content and ways of receiving the information.
In Q methodological studies, data are collected in the form of *Q sorts* that represent a participant’s subjective viewpoint. Each participant creates a personal *Q sort* by ranking a set of statements covering different aspects of the studied phenomenon (i.e. *Q set*) along a standardised ranking distribution or sorting grid (Figure 1). The *Q set* is sampled from the *concourse*, which is a collection of experience and knowledge representing different views and opinions on the phenomenon of interest.\[161\] After *Q sorts* have been collected, intercorrelations between *Q sorts* are analysed with Q factor analysis.\[160,162\]

In Paper V, the *Q set* (i.e. the set of statements) was drafted from the *concourse*, based on the interviews in Paper IV, the current peer-reviewed literature, and communications with mothers of children having DS. Through discussions in the research group (with clinical experience as an obstetrician, midwife and genetic counsellor), the statements covering different aspects of information about PND and CAs were reduced and refined. The final *Q set* consisted of 50 statements (displayed in Table 6 in the results section), which the participants were asked to rank in the fixed sorting grid (Figure 1), with ranking values ranging from +5 (most important) to -5 (least important), based on the question “What is important to you when you receive information about PND?”. Participants were asked to give written comments on their sorting process, resulting in some qualitative data.

![Figure 1. The fixed quasi-normal distribution sorting grid employed for Paper V. Participants sorted the 50 items (i.e. statements) of the Q set, one item per square in the grid, to indicate their prioritisation of the items relative to each other. For example, only 2 items can be ranked as least important at -5.](image)

Two questionnaires were utilised, in addition to the *Q set*; a questionnaire developed specifically for the study covering sociodemographic and background characteristics, and the validated “Threatening Medical Situations Inventory (TMSI)” questionnaire to identify coping styles.\[163\]
Pregnant women were invited to participate in the study by a midwife when they called to schedule their first appointment at an outpatient antenatal care unit in Uppsala, Örebro or Eskilstuna between January 2020 and October 2021. If interested, they were sent written information about the study, together with the Q set (50 cards, one card for each statement) and the two questionnaires. Participants were instructed to fill in the questionnaires and sort the set of cards at home and return them by post. Inclusion criteria were women in early pregnancy, Swedish speaking and aged 18 or above. Some participants waited to fill in the questionnaires and completing the Q sort, therefore, some had received information about PND and some had undergone PND when they completed and returned the study material. Fifty pregnant women were initially included, but as intermediate Q factor analysis indicated saturation at 45 participants, the in-depth factor interpretation was performed with this subset.

Ethical considerations
All studies included in this thesis were approved by the Regional Ethical Review Board at the Medical Faculty of Uppsala University in Sweden (Paper I-II: 2010/403, Paper III: 2010/403/1, Paper IV: 2015/227, Paper V: 2019-00267). Due to difficulties in recruitment for study IV, adjustments were made to the recruitment method. Supplementary applications were approved by the same Regional Ethical Review Board (2015/227/1, 2015/227/2, 2015/227/3).

Participation in the studies was optional and did not affect the care provided to the expectant parents in Papers I, II, IV and V. Questionnaires (Paper I-III) were collected anonymously, ensuring no possibility of identification. Answering the questionnaire was regarded as equivalent to informed consent. In Paper IV, participants received both written and verbal information about the study at a separate occasion prior to the interviews. Verbal consent was obtained before starting the interview. The participants in Paper V received brief verbal information, followed by written information about the study, after which written consent was collected. Participants across all papers were given contact information for questions regarding the studies.

Analyses
Quantitative statistical analyses (Papers I-III)
Statistical analyses for Papers I-III were performed using the Statistical Package for the Social Sciences (SPSS) version 22 (IBM Corp., Armonk, NY, USA). A significance level of 0.05 was applied throughout. Descriptive analyses were performed to present demographic variables. In all questionnaires
one of the response alternatives for the multiple-choice questions regarding knowledge about DS was predetermined as correct. When these knowledge questions were analysed, the responses were dichotomised into correct and incorrect. A two-tailed Chi-square test was used to calculate differences in knowledge (correct vs. incorrect) between women and partners and by demographic background, between women accepting invasive tests and women declining PND, and between midwives with varying occupational experience. For women who answered the knowledge questions incorrectly in Paper II, differences in overestimation and underestimation between the accepting and declining groups were also calculated using the Chi-square test. The independent two-sided t-test was used for other comparisons across Papers I-III.

Qualitative content analysis (Paper IV)

The interviews were recorded, transcribed verbatim in Swedish and then analysed using qualitative content analysis. To ensure that no nuances or meanings were lost in translation, all analyses were conducted in Swedish. Initially, the transcripts were read through several times to get a sense of the whole. The qualitative data software tool Nvivo (version 11.4.3), was used to organise the qualitative data for analysis. Meaning units relevant to the decision-making process regarding PND were identified. These meaning units were condensed and abstracted into codes. In the next step, the codes were sorted into subcategories and categories, eventually resulting in themes. An inductive approach to content analysis was applied, i.e. themes and categories were not identified in advance but emerged from the coding process. The first author was responsible for the analysis, but results were discussed continuously with the research team to reach consensus on the interpretation. The quotes used in Paper IV were translated into English by a professional translator.

Q factor analysis (Paper V)

The Q dedicated software PQMethod (version 2.35), specifically developed for analysing data from Q-methodological studies, was used. It computes inter-correlations among the participants’ Q sorts and subsequently conducts factor-analysis. Q factor analysis usually generates a number of groups, i.e. factors, with similarly sorted statements (Q set). These factors represent the shared viewpoints among the Q-sorts that have sorted the Q set (i.e. statements) similarly. Hence, different viewpoints on a subject are condensed into factors, which represent ways of thinking (subjective views). An inductive approach to Q factor analysis with Centroid factor extraction and Varimax factor rotation was used. At first, seven factors were extracted. However, the factor loadings and eigenvalues of the seven
factors indicated that a three-factor solution was appropriate based on the Kaiser-Guttman criterion, Humphrey’s rule and factors with two or more significant loadings.\textsuperscript{160} Factor extraction was therefore repeated with three factors, followed by Varimax factor rotation, where defining \textit{Q sorts} (unique loaders) for each factor were identified using the auto flag option in PQMethod. These \textit{defining Q sorts} are the individual \textit{Q sorts} produced by participants who share the viewpoint of one factor. Through weighted averages of the defining \textit{Q sorts}, PQMethod then calculates an \textit{idealised Q sort} for each of the three factors, which are displayed in a \textit{factor array} (Table 6). A person who shares the exact same viewpoint as a factor would sort the items like the \textit{idealised Q sort} of that factor. To aid in the factor interpretation process, Crib sheets\textsuperscript{160} were produced based on the factor arrays.

Intermediate factor analyses were performed with 30, 40, 45 and 50 participants. When comparing intermediate analyses between 45 and 50 participants, the background characteristics, factor loadings and factor arrays indicated that the participant group was sufficiently heterogeneous, and that saturation was reached at 45 participants. Accordingly, recruitment was discontinued, and in-depth factor interpretation was performed with 45 participants. This factor interpretation was based on factor arrays, crib sheets,\textsuperscript{160} distinguishing statements (i.e. statements that were significantly differently ranked between factors), \textit{consensus statements} (i.e. statements that did not distinguish between any pair of factors) at a \(p < 0.01\) level, as well as qualitative written comments on the sorting process and sociodemographic data from the questionnaire.

Coping styles were analysed by summing up the relevant items for monitoring and blunting in each scenario within the TMSI questionnaire, resulting in a total blunter score and a total monitoring score. By subtracting the blunter scores from the monitor scores, a total score was obtained, where lower (negative) scores indicated a blunting coping style, and higher scores indicated a monitoring coping style.

Comparisons among the three viewpoints regarding sociodemographic data and coping styles were performed with Fisher-Freeman-Halton exact test (for comparisons of nominal data) and Independent-Samples Kruskal-Wallis test (for comparisons of continuous ratio data (i.e. age)), using SPSS version 28.0.1. A significance level of \(p < 0.05\) was applied for all comparisons. To compare women with or without personal experience of someone with a congenital anomaly, the response alternatives were sorted into ‘yes’ and ‘no’ due to small numbers, where ‘yes’ includes the participants choosing any of the yes alternatives (within the close family, extended family and/or among friends).
Results

The results are presented separately for each paper. In addition, for Papers I and II some of the results are presented together to allow for comparisons among the three participant groups.

Papers I and II

Demographics and comparisons for Papers I and II

Demographic information on the women in all three groups for Papers I and II (accepting CUB testing, accepting invasive testing and declining PND for DS) is presented in Table 2. The mean age was 33 years for women in the CUB group, 35 years for the invasive group, and 37 years for the declining PND group.

Table 2. Demographic data concerning the women in Papers I and II.

<table>
<thead>
<tr>
<th></th>
<th>CUB test n = 105</th>
<th>Invasive test n = 76</th>
<th>Declining PND n = 65</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age ≥ 35 years</td>
<td>46/44</td>
<td>45/59</td>
<td>63/97</td>
</tr>
<tr>
<td>Residence</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Suburban</td>
<td>33/31</td>
<td>21/28</td>
<td>24/37</td>
</tr>
<tr>
<td>Urban</td>
<td>72/69</td>
<td>54/71</td>
<td>40/62</td>
</tr>
<tr>
<td>Co-habiting</td>
<td>103/98</td>
<td>71/93</td>
<td>64/99</td>
</tr>
<tr>
<td>Highest achieved educational level</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Elementary school</td>
<td>1/1</td>
<td>3/4</td>
<td>1/2</td>
</tr>
<tr>
<td>High school</td>
<td>30/29</td>
<td>22/29</td>
<td>17/26</td>
</tr>
<tr>
<td>University</td>
<td>73/70</td>
<td>50/66</td>
<td>47/72</td>
</tr>
<tr>
<td>Previous children</td>
<td>56/53</td>
<td>47/62</td>
<td>54/83</td>
</tr>
<tr>
<td>CUB test in previous pregnancy*</td>
<td>21/20</td>
<td>-/ -</td>
<td>1/2</td>
</tr>
<tr>
<td>Invasive test in previous pregnancy**</td>
<td>-/ -</td>
<td>75/99</td>
<td>-/ -</td>
</tr>
</tbody>
</table>

Missing values varied between 0-3.8 (with the exception of * and **) for the questions on CUB test in previous pregnancy for the CUB group; residence, previous children and invasive test in a previous pregnancy for the invasive group; residence and CUB test in a previous pregnancy for the declining group.

*Missing values in the invasive group were 98.7%.

**Missing values in the CUB group and the declining group were 100%.
Across all three groups studied in Papers I and II, we found varying, and in many respects low levels of knowledge about cognitive, medical and social consequences of DS (Figures 2-4). When comparing pregnant women in the CUB group with those in the invasive group, there were no significant differences in knowledge of DS. However, when comparing women in the invasive test group with the declining group, a significant difference was found in just one knowledge question: walking ability (p=0.03). This difference remained when comparing all women undergoing PND (CUB and invasive test) to women declining PND (p = 0.02). In addition, a significant difference was also seen for another question: reading ability (p = 0.04). For both these questions more women who accepted PND answered correctly.

Regarding participants who had someone with DS in their vicinity, there was a significant difference between groups. More women declining PND (28%) knew someone with DS compared to women choosing to undergo invasive tests (13%) (p =0.03). The women in the CUB group had similar numbers (16%) to the invasive group, and the significant difference remained when comparing all women undergoing PND (CUB and invasive tests) to women declining PND (p = 0.02).

Figure 2. Knowledge about cognitive consequences. The percentage of expectant parents who answered correctly on questions concerning the percentage of children with Down syndrome who learn to walk, talk and read. Four response alternatives were given; 0%, 20%, 50% and 100%. The correct answers were: 100% for walking ability, 50% for talking ability and 20% for reading ability. The non-response rate varied between 1.0-4.6%.
Figure 3. Knowledge about medical consequences. The percentage of expectant parents who answered correctly to questions concerning how many children with DS who have heart anomalies, gastrointestinal anomalies and hearing problems. Four response alternatives were given: 10%, 50%, 70% and 100%, where the correct answers were 50% for heart anomalies, 10% for gastrointestinal anomalies and 70% for hearing problems. The non-response rate varied between 1.9-7.7%.

Figure 4. Knowledge about social consequences. The percentage of expectant parents who answered correctly on questions concerning differences between families with or without a child with DS concerning divorces, number of siblings, living with biological parents and parents working outside home. Three response alternatives were given: ‘Fewer’, ‘Same’ and ‘More’. The correct answers were ‘Same’ for all four questions. The non-response rate varied between 1.0-3.9%.
In table 3, aspects of information, such as informational sources, received information and satisfaction with information are shown for all three groups in Papers I and II. These results are also discussed separately for each paper below.

Table 3. Information about PND and DS for expectant parents undergoing a CUB test (women and partners n = 209), women undergoing invasive tests (n = 76) and women declining PND for CAs (n =65).

<table>
<thead>
<tr>
<th>Source of information about PND*</th>
<th>CUB test (N, %)</th>
<th>Invasive test (N, %)</th>
<th>Declining PND (N, %)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Midwife</td>
<td>80 (38.3)</td>
<td>20 (26.3)</td>
<td>46 (70.8)</td>
</tr>
<tr>
<td>Doctor</td>
<td>1 (0.5)</td>
<td>12 (15.8)</td>
<td>0 (0)</td>
</tr>
<tr>
<td>Internet</td>
<td>26 (12.4)</td>
<td>17 (22.4)</td>
<td>4 (6.2)</td>
</tr>
<tr>
<td>Friends</td>
<td>14 (6.7)</td>
<td>3 (3.9)</td>
<td>0 (0)</td>
</tr>
<tr>
<td>Several sources</td>
<td>63 (30.1)</td>
<td>18 (23.7)</td>
<td>13 (20.0)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Source of information about DS*</th>
<th>CUB test (N, %)</th>
<th>Invasive test (N, %)</th>
<th>Declining PND (N, %)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Midwife</td>
<td>41 (19.6)</td>
<td>6 (7.9)</td>
<td>22 (33.8)</td>
</tr>
<tr>
<td>Doctor</td>
<td>1 (0.5)</td>
<td>6 (7.9)</td>
<td>0 (0)</td>
</tr>
<tr>
<td>Internet</td>
<td>56 (26.8)</td>
<td>30 (39.5)</td>
<td>12 (18.5)</td>
</tr>
<tr>
<td>Friends</td>
<td>30 (14.4)</td>
<td>11 (14.5)</td>
<td>14 (21.5)</td>
</tr>
<tr>
<td>Several sources</td>
<td>35 (16.7)</td>
<td>6 (7.9)</td>
<td>4 (6.2)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Received sufficient information about</th>
<th>CUB test (N, %)</th>
<th>Invasive test (N, %)</th>
<th>Declining PND (N, %)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal tests in general</td>
<td>167 (79.9)</td>
<td>61 (80.3)</td>
<td>52 (80.0)</td>
</tr>
<tr>
<td>Prenatal tests for DS</td>
<td>157 (75.1)</td>
<td>51 (67.1)</td>
<td>47 (72.3)</td>
</tr>
<tr>
<td>Living with a child with DS</td>
<td>49 (23.4)</td>
<td>15 (19.7)</td>
<td>22 (33.8)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Satisfied with information regarding</th>
<th>CUB test (N, %)</th>
<th>Invasive test (N, %)</th>
<th>Declining PND (N, %)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Content</td>
<td>147 (70.3)</td>
<td>52 (68.4)</td>
<td>53 (81.5)</td>
</tr>
<tr>
<td>How it was provided</td>
<td>143 (68.4)</td>
<td>46 (60.5)</td>
<td>53 (81.5)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Would like to know more about DS**</th>
<th>CUB test (N, %)</th>
<th>Invasive test (N, %)</th>
<th>Declining PND (N, %)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>130 (62.2)</td>
<td>40 (52.6)</td>
<td>-</td>
</tr>
</tbody>
</table>

Missing values varied between 1.5%-14.5%.
*Other informational sources (not displayed in the table) were stated as written comments by 1.5%-15.3%.
**Women declining PND were not asked if they wanted more information about DS

Table 4 provides an overview of reasons to accept or decline PND and includes answers from women undergoing CUB (Paper I) and invasive testing as well as women declining PND for CAs (Paper II).
Table 4. Reasons to undergo or abstain from PND for women undergoing a CUB test (n = 105), invasive tests (n = 76) and declining PND for CAs (n = 65). Not all response alternatives were applicable for all groups (marked with - in the table). Several response alternatives could be given.

<table>
<thead>
<tr>
<th>Reasons to undergo CUB or invasive testing?</th>
<th>CUB N (%)</th>
<th>Invasive N (%)</th>
<th>Declining N (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Because you should do it if older than 35*</td>
<td>25 (23.8)</td>
<td>18 (23.7)</td>
<td>-</td>
</tr>
<tr>
<td>To perform all tests possible to make sure the baby is healthy</td>
<td>70 (66.7)</td>
<td>34 (44.7)</td>
<td>-</td>
</tr>
<tr>
<td>To ease my worries</td>
<td>58 (55.2)</td>
<td>48 (63.2)</td>
<td>-</td>
</tr>
<tr>
<td>Because I/my partner have/has a child with an illness</td>
<td>2 (1.9)</td>
<td>5 (6.6)</td>
<td>-</td>
</tr>
<tr>
<td>Because my partner wanted to</td>
<td>11 (10.5)</td>
<td>6 (7.9)</td>
<td>-</td>
</tr>
<tr>
<td>Screening indicated high risk</td>
<td>-</td>
<td>10 (13.2)</td>
<td>-</td>
</tr>
<tr>
<td>Because of the cost of CUB testing</td>
<td>-</td>
<td>1 (1.3)</td>
<td>-</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Reasons to abstain from PND**</th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Termination of pregnancy is not an option</td>
<td>-</td>
<td>-</td>
<td>38 (58.5)</td>
</tr>
<tr>
<td>I can take care of a child with DS</td>
<td>-</td>
<td>-</td>
<td>28 (43.1)</td>
</tr>
<tr>
<td>I have a low risk</td>
<td>-</td>
<td>-</td>
<td>19 (29.2)</td>
</tr>
<tr>
<td>DS is not a serious condition</td>
<td>-</td>
<td>-</td>
<td>23 (35.4)</td>
</tr>
<tr>
<td>Invasive testing increases the risk of miscarriage</td>
<td>-</td>
<td>-</td>
<td>37 (56.9)</td>
</tr>
<tr>
<td>Because of the cost</td>
<td>-</td>
<td>-</td>
<td>1 (1.5)</td>
</tr>
</tbody>
</table>

*The numbers in the table concern all women undergoing CUB or invasive tests. Of women ≥35 years, 48% in the CUB group and 40% in the invasive group stated this reason to have a test.

**Two participants did not answer this question (missing value 3%)

Paper I

When comparing women and partners undergoing a CUB test, there were few differences. However, there was one significant difference regarding knowledge, concerning the question about walking ability (p = 0.006). Other differences found concerned reasons for choosing to undergo a CUB test. Significantly more women (55%) than partners (33%) chose the test because of worry (p = 0.009). Conversely, more partners (24%) than women (11%) chose the test because their spouse wanted to (p = 0.003). Women’s reasons to undergo CUB are displayed in Table 4. Since differences were few, other results are presented for the whole group of expectant parents in the CUB group (women + partners) below unless otherwise stated.

The most commonly reported main sources of information about PND were midwives, but many reported using several informational sources (Table 3). For information about DS, the Internet was the most frequently reported informational source. The majority of expectant parents had received information about prenatal tests for DS, but only 23% had information about what it means to live with a child with DS. Most were satisfied with the content of
the information and how it was given, although many wanted more information about DS.

When asked what they would do if the CUB test indicated an increased probability of DS, 24% of all expectant parents stated that they had not yet decided if they wanted to undergo an invasive test. If DS was diagnosed after a diagnostic test, 46% had not decided what to do, and 43% thought they would terminate the pregnancy.

Regarding knowledge about mean survival age for individuals with DS, 53% of the women and 43% of the partners in the CUB group answered correctly, where 60 years was chosen as the correct value. Women’s and partner’s answers to other knowledge questions are presented in Figures 2-4.

Paper II
Aspects on information are displayed in Table 3. The most frequently reported main source of information about PND for women accepting invasive tests was a healthcare professional or the Internet. For women declining PND, midwives were the single most commonly reported source. Women in both groups reported using several informational sources. Regarding information about DS, the accepting group reported the Internet as the most common informational source, but some received information from friends or healthcare professionals. In the declining group, midwives remained the most common source, followed by friends and the Internet.

Similarly to the CUB group, a majority of pregnant women, both accepting invasive testing and declining PND for DS, felt they had received sufficient information about prenatal tests for DS and were satisfied with the content as well as how it was given. When comparing the two groups, significantly more women declining PND were satisfied with the information (p = 0.01) and how it was given (p = 0.01). Significantly more women who declined PND reported feeling they had received sufficient and adequate information about what it entails to raise a child with DS (p = 0.02), compared with women who chose invasive PND. More information about DS was desired by half of women undergoing invasive PND.

Reasons to undergo or abstain from PND are displayed in Table 4. The most common reasons for accepting invasive PND were worry, wish for reassurance of a healthy baby and age (>35 years). Other reasons stated in comments included having DS or a sick child in the family or a sick child in a previous pregnancy. The most common reasons for declining PND were not regarding TOP as an option, feeling they could take care of a disabled child and the risk of miscarriage. Other reasons mentioned in comments included ethical concerns and not knowing what to do with the information if the test indicated DS. When women undergoing invasive testing were asked what they would do if DS was diagnosed, 21% had not decided what to do, 74% thought
they would terminate the pregnancy, and 5% would prepare to take care of the child.

Regarding knowledge of DS, 50% of the women in the invasive group and 52% of the women in the declining group knew the average survival age for people with DS. Figures 2-4 depict their responses to the other knowledge questions.

**Paper III**

The midwives included in Paper III were all female, with a mean age of 50 years (range 31-64 years). The year of graduation varied between 1974 and 2012, with varying years of experience working as a midwife (1-38 years).

Midwives had varying, and in some respects low levels of knowledge about DS and its medical, cognitive and social aspects (Table 5). The talking and reading abilities of individuals with DS were overestimated, while hearing problems were largely underestimated. Questions regarding social aspects were correctly answered by more than half of the participants, however, a substantial minority overestimated both the number of siblings and parents’ divorce rates. When comparing those who had worked less than 10 years to those who had worked more than 10 years, there were no significant differences in knowledge or in the information they provided to expectant parents.

Table 5. The midwives’ knowledge of DS and the extent to which they overestimated or underestimated when answering incorrectly.

<table>
<thead>
<tr>
<th>Knowledge questions</th>
<th>Underestimate</th>
<th>Correct answer</th>
<th>Overestimate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mean survival age (60 years)</td>
<td>19</td>
<td>42</td>
<td>1</td>
</tr>
<tr>
<td>How many learn to walk (100%)</td>
<td>2</td>
<td>61</td>
<td>0</td>
</tr>
<tr>
<td>How many learn to talk (50%)</td>
<td>1</td>
<td>22</td>
<td>36</td>
</tr>
<tr>
<td>How many learn to read (20%)</td>
<td>0</td>
<td>18</td>
<td>40</td>
</tr>
<tr>
<td>Frequency of cardiac anomalies (50%)</td>
<td>7</td>
<td>44</td>
<td>13</td>
</tr>
<tr>
<td>Frequency of gastrointestinal anomalies (10%)</td>
<td>0</td>
<td>46</td>
<td>17</td>
</tr>
<tr>
<td>Frequency of hearing problems (70%)</td>
<td>55</td>
<td>8</td>
<td>0</td>
</tr>
<tr>
<td>How many live with biological parents (&gt;90%)</td>
<td>17</td>
<td>46</td>
<td>0</td>
</tr>
<tr>
<td>Number of siblings (same)</td>
<td>8</td>
<td>36</td>
<td>16</td>
</tr>
<tr>
<td>Divorce rate for parents (same)</td>
<td>7</td>
<td>34</td>
<td>22</td>
</tr>
<tr>
<td>Frequency of parents working (same)</td>
<td>16</td>
<td>46</td>
<td>1</td>
</tr>
</tbody>
</table>

Non-response rates varied between 0 and 9.4%.

Information about second-trimester ultrasound and CUB was provided to all expectant parents by a majority of the midwives (94% and 73%, respectively). Information about invasive tests (36%) and DS (20%) were provided less frequently to all expectant parents, and information about DS was mostly given only when requested or when there was an increased probability of DS.
majority felt they had complete or partial knowledge to inform expectant parents about second-trimester ultrasound (99%), CUB (83%) and invasive tests (80%). Almost half of the participants did not feel they had sufficient knowledge to inform about DS, and 9% felt they had completely sufficient knowledge about DS. Occupational experience had a significant impact on their self-perceived knowledge, and midwives with more than 10 years of working experience more often felt competent in giving information about invasive tests (p = 0.028) and DS (p = 0.007).

The most frequently asked questions from expectant parents were, according to the midwives, those concerning the PND methods regarding possible risks (78%), when they could be performed (63%), and what could be detected (31%). While one-third (31%) of the midwives had observed an increasing interest in DS with more questions from expectant parents in the last 3 years, no one reported questions about DS and its consequences as most frequently asked. Questions about DS were mostly reported coming from older women (70%), and the most common questions concerned the effect of maternal age on the probability of a DS pregnancy (69%) and the incidence of DS (53%). Symptoms of DS were regarded as the most commonly asked questions concerning DS by one midwife.

Few midwives reported having received sufficient education regarding ultrasound (11%), CUB (17%), invasive tests (11%) and DS (3%). A majority (94%) desired more education in one or more of the mentioned areas.

Paper IV

A total of 24 pregnant women (gestational weeks 6-11) were interviewed before undergoing PND. At the time of the interview, a majority (21/24) had not yet met with a midwife and thus had not received information about PND. None had undergone previous PND (except for first- and/or second-trimester ultrasound in a previous pregnancy). The participants were aged between 25 and 36 years, had diverse sociodemographic backgrounds with varying educational levels (58% college or university education) and parity (63% nulliparous). Almost half had no previous personal experience of anomalies, while the rest had experience of various conditions from family, friends or work. Two main themes were found to influence the decision-making process regarding PND: Individual factors and External factors. An overview of the themes, their categories and subcategories are displayed in Figure 5.
Individual factors – The women’s experiences, perceptions and values

The decision-making process regarding PND is influenced by individual factors. Women’s attitudes towards anomalies affect their decision, which, in turn, can be influenced by previous experiences of anomalies from family, friends and work. For some, having PND can be a way to be able to prepare for the birth of a child with an anomaly. Others may consider having a TOP in case of an anomaly and undergo PND to gain as much information as possible about the fetus. For these women, what conditions a test can detect and their assessment of how the condition will affect the child and the family are often major factors affecting the decision-making regarding PND.

Worry and need for reassurance can be another reason to undergo PND to get confirmation of the pregnancy and the health of the baby. The wish to have a first-trimester ultrasound was to some a reason to decide to undergo CUB.
However, feelings of uncertainty related to testing, such as that of a non-diagnostic probability assessment, like the CUB test, and not knowing how to handle the results, may be reasons to abstain from testing. Especially for women who know in advance that they will not have an invasive test due to the associated risks, uncertainty may motivate abstaining from screening.

Some women described their self-perceived risk of expecting a fetus with a CA or other anomaly as an important factor affecting their perceived need to have PND or not. Many women were aware that the probability of CAs increases with increasing maternal age. Thus, valuation of age was important when assessing the self-perceived risk. Some women also mentioned the impact of genetic or other predisposing factors that may increase the probability of a CA as something that they would consider in their decision-making. Women’s self-perceived risk was influenced by their perceptions of healthcare professionals’ assessment of risk. Some women assumed that the midwife made a professional assessment of their individual need to have PND and expected to be given such information, which would motivate having tests. Not being offered or recommended a test was perceived, to some, as a confirmation that the midwife had assessed their individual probability as low and testing was not necessary.

External factors – The women’s impression of the test and others’ views

The decision-making process regarding PND is also influenced by external factors, such as test characteristics, where the most influential seems to be test-related risks that often times were deal-breakers for women. Test accuracy could also influence the decision, as some women were not interested in taking a test that did not provide a clear answer. Results from a probability assessment, like CUB, were difficult to interpret. Especially for women who would not consider invasive tests due to the miscarriage risk, the decision-making regarding screening tests was more complicated than for women who were open to having an invasive test to verify a diagnosis. The timing of the test and waiting time for results also mattered, both for women who could consider TOP in case of an anomaly and therefore had a time limit, as well as for women who sought to alleviate their worries with PND and therefore preferred early testing and short waiting times. Some women also felt it would be emotionally harder to have a TOP later in the pregnancy.
Women also described influence from others, many times influence from their partner, as many couples strive for a joint decision regarding the pregnancy. They expressed a wish to have time to discuss and reflect with their partner after having received information about PND, before making a decision together. Healthcare professionals' attitudes towards PND were described as potentially affecting women in either direction, to have or to abstain from PND. Some women described feeling pressured to have a test if healthcare professionals presented it as something you should do, while others felt that a judgemental attitude from healthcare professionals towards PND and TOP could influence them not to have the test. The organisation of healthcare services and an uneven offer of PND, i.e. CUB at a subsidised cost to specific age groups (e.g. >35), hampered the possibility of having PND for younger women who would otherwise like to have a CUB test. In contrast, women over 35 sometimes felt compelled to have PND since the age limit indirectly emphasised their increased probability of having a CA, and for these women, the offer could feel like a recommendation from the healthcare.

Paper V

A total of 45 pregnant women from various sociodemographic backgrounds from three middle-sized cities (Uppsala (n = 17), Örebro (n = 7) and Eskilstuna (n = 21)) were included. The mean age was 29.9 (range 23-38), with a majority having higher education (72% university /college) and almost half were parous. Three factors were found that represent different viewpoints on what is important to pregnant women when receiving information about PND. Together, these three factors explain 56% of the study variance. Each viewpoint consists of Q sorts of participants who share that viewpoint (i.e. defining Q sorts), with 15 Q sorts in factor 1, 8 in factor 2, and 11 in factor 3. No significant differences were found between the participants in the three viewpoints when comparing their background demographics (e.g. city, age, parity, education, marital status, religion, experience of anomalies, attitudes towards PND and TOP) and coping styles.

Table 6 provides an overview of the three factors with a factor array that includes all the 50 statements and the ranking of the statements for each factor (i.e. exemplar Q sorts). The factor array also includes which statements are consensus statements, i.e. did not distinguish between any pair of factors (n =17) or distinguishing, i.e. statements that were ranked significantly differently between factors (n = 32). A statement can distinguish one factor from
the others or all factors from each other, and the distinguishing statements, therefore, indicate which factor they are distinguishing for (F1, F2, F3).

All viewpoints considered receiving information from someone with sufficient knowledge as most important. In general, women prioritised having their thoughts and questions taken seriously, as well as having time to reflect between receiving the information and making decisions. Conversely, the least important aspect was to have the opportunity to discuss religious values related to PND and to receive verbal information in a group setting. The viewpoints of the factors are described below.

**Factor 1: Stepwise information and decision-making**

Women with viewpoint 1 perceive information and decision-making as a step-by-step process. In step 1, they prioritise information and decision-making regarding PND. If screening indicates an anomaly, they wish to receive information about the conditions screened for as step 2. They do not want to receive too much information on one occasion or too early in the pregnancy. It is important to them that their partner is present when receiving information, but not as important for them to be involved in the decision-making process.

**Factor 2: Decision-making as a continuous process based on couple autonomy**

Women of viewpoint 2 strive for an informed decision as a couple about the complete concept of accepting or declining PND and how to handle the test results. They value receiving information about test methods and about various aspects of the conditions tested for. They wish to receive and make decisions together with their partner. They value having the opportunity to discuss ethical values related to PND, and to have time for reflection before making a decision.

**Factor 3: As much information as early as possible – The importance of personal autonomy in decision-making**

Women of viewpoint 3 prioritise autonomous decision-making based on non-directive information early in the pregnancy. They wish to receive extensive information, especially about test methods, and also about the conditions tested for. They value the meeting with healthcare professionals and having sufficient time with them, but they do not wish to discuss ethical values related to PND. They prioritise autonomous decision-making and have less need to involve a partner in the process.
Table 6. Factor array with the 50 statements and the ranking of each statement for the three factors: Factor 1 (F1), Factor 2 (F2), Factor 3 (F3).

<table>
<thead>
<tr>
<th>No.</th>
<th>Statement</th>
<th>F1</th>
<th>F2</th>
<th>F3</th>
<th>Distinguishing or consensus</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>That the person providing me the information has sufficient knowledge on the subject.</td>
<td>5</td>
<td>5</td>
<td>3</td>
<td>Consensus</td>
</tr>
<tr>
<td>2</td>
<td>That both my partner and I receive information about prenatal diagnosis.</td>
<td>3</td>
<td>3</td>
<td>3</td>
<td>Consensus</td>
</tr>
<tr>
<td>3</td>
<td>To receive information about possible choices regarding pregnancy management if the test indicates an anomaly (further tests, continued management during pregnancy or a possible termination of pregnancy).</td>
<td>3</td>
<td>2</td>
<td>3</td>
<td>Consensus</td>
</tr>
<tr>
<td>4</td>
<td>To emphasize that prenatal diagnosis is not a guarantee to have a healthy baby.</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>Consensus</td>
</tr>
<tr>
<td>5</td>
<td>To receive neutral information that does not reflect the personal values of the midwife/doctor.</td>
<td>4</td>
<td>2</td>
<td>3</td>
<td>F1</td>
</tr>
<tr>
<td>6</td>
<td>To receive oral information individually.</td>
<td>0</td>
<td>-3</td>
<td>0</td>
<td>F2</td>
</tr>
<tr>
<td>7</td>
<td>To receive information on the reliability of a test and its strengths and limitations.</td>
<td>3</td>
<td>2</td>
<td>3</td>
<td>Consensus</td>
</tr>
<tr>
<td>8</td>
<td>That both my partner and I have the possibility to be part of the decision to undergo or abstain prenatal diagnosis.</td>
<td>1</td>
<td>4</td>
<td>-2</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>9</td>
<td>To receive information on prenatal diagnosis at a separate occasion when no other pregnancy related information is given.</td>
<td>-1</td>
<td>-4</td>
<td>-4</td>
<td>F1</td>
</tr>
<tr>
<td>10</td>
<td>To receive equal and corresponding information about all the chromosomal anomalies (trisomy 13, 18, 21) that prenatal diagnosis is offered for, even if Down syndrome (trisomy 21) is the most common chromosomal condition.</td>
<td>1</td>
<td>-1</td>
<td>1</td>
<td>F2</td>
</tr>
<tr>
<td>11</td>
<td>To not receive too much information at one occasion.</td>
<td>0</td>
<td>-2</td>
<td>-3</td>
<td>F1</td>
</tr>
<tr>
<td>12</td>
<td>That it is emphasized that information about prenatal diagnosis is optional.</td>
<td>0</td>
<td>1</td>
<td>-3</td>
<td>F3</td>
</tr>
<tr>
<td>13</td>
<td>That the information is given early in the pregnancy.</td>
<td>2</td>
<td>0</td>
<td>4</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>14</td>
<td>To have the possibility to discuss religious values related to prenatal diagnosis.</td>
<td>-3</td>
<td>-3</td>
<td>-5</td>
<td>Consensus</td>
</tr>
<tr>
<td>15</td>
<td>To receive information that Down syndrome leads to intellectual disability to a varying degree.</td>
<td>-1</td>
<td>1</td>
<td>0</td>
<td>Neither</td>
</tr>
<tr>
<td>16</td>
<td>To receive some information on the conditions that can be detected, e.g. Down syndrome before I make a decision whether to undergo or abstain prenatal diagnosis.</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>Consensus</td>
</tr>
<tr>
<td>17</td>
<td>To have the possibility to discuss ethical values related to prenatal diagnosis.</td>
<td>-1</td>
<td>0</td>
<td>4</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>18</td>
<td>To receive as much information as possible.</td>
<td>1</td>
<td>2</td>
<td>4</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>19</td>
<td>That the incidence of the medical consequences among children with Down syndrome is put in relation to the incidence among other children.</td>
<td>-1</td>
<td>-2</td>
<td>-3</td>
<td>Consensus</td>
</tr>
<tr>
<td>20</td>
<td>To receive information about when in the pregnancy different tests can be done.</td>
<td>2</td>
<td>0</td>
<td>2</td>
<td>F2</td>
</tr>
<tr>
<td>21</td>
<td>That the midwife/doctor have sufficient time for me.</td>
<td>2</td>
<td>4</td>
<td>5</td>
<td>F1</td>
</tr>
<tr>
<td>22</td>
<td>To receive information on how family life, the parent’s relationship or siblings can be affected by having a child with Down syndrome in the family.</td>
<td>-2</td>
<td>1</td>
<td>-1</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>23</td>
<td>To receive information about the limitations of the tests, i.e. what the test cannot detect.</td>
<td>1</td>
<td>0</td>
<td>2</td>
<td>F2</td>
</tr>
<tr>
<td>24</td>
<td>To receive information on special demands on me when caring for a child with Down syndrome.</td>
<td>-2</td>
<td>1</td>
<td>1</td>
<td>F1</td>
</tr>
<tr>
<td>25</td>
<td>To receive information on care programs and available support from the society for children with Down syndrome and their families.</td>
<td>-3</td>
<td>0</td>
<td>0</td>
<td>F1</td>
</tr>
<tr>
<td>26</td>
<td>To receive information on risks of the different tests.</td>
<td>-4</td>
<td>-2</td>
<td>3</td>
<td>Consensus</td>
</tr>
<tr>
<td>27</td>
<td>That I can make a decision about prenatal diagnosis without pressure from the midwife/doctor.</td>
<td>-1</td>
<td>3</td>
<td>3</td>
<td>F2</td>
</tr>
<tr>
<td>28</td>
<td>To get advice from my midwife/doctor on whether to have a test or not.</td>
<td>0</td>
<td>-3</td>
<td>-2</td>
<td>F1</td>
</tr>
<tr>
<td>29</td>
<td>To receive information that it is not possible to foresee how severe the symptoms will be for a specific individual with Down syndrome just by looking at the chromosomess.</td>
<td>-1</td>
<td>-1</td>
<td>-1</td>
<td>Consensus</td>
</tr>
<tr>
<td>30</td>
<td>To receive information also about tests that are not offered in my region, but are offered in other regions or by private clinics.</td>
<td>-1</td>
<td>0</td>
<td>0</td>
<td>Consensus</td>
</tr>
<tr>
<td>31</td>
<td>That my questions/thoughts are taken seriously.</td>
<td>0</td>
<td>3</td>
<td>4</td>
<td>Consensus</td>
</tr>
<tr>
<td>32</td>
<td>To receive information about what life can look like for a child with Down syndrome during childhood, school years and adolescence.</td>
<td>-3</td>
<td>1</td>
<td>-1</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>33</td>
<td>To have time to reflect between receiving information and making a decision.</td>
<td>1</td>
<td>3</td>
<td>2</td>
<td>Consensus</td>
</tr>
<tr>
<td>34</td>
<td>To receive information about the cognitive consequences of Down syndrome, e.g. the ability to talk, read, write or walk.</td>
<td>-4</td>
<td>1</td>
<td>-1</td>
<td>F1</td>
</tr>
<tr>
<td>35</td>
<td>To receive information about the medical consequences of Down syndrome (e.g. congenital heart defects, hearing problems, growth retardation, abnormalities).</td>
<td>-2</td>
<td>0</td>
<td>0</td>
<td>F1</td>
</tr>
<tr>
<td>36</td>
<td>To receive written information.</td>
<td>0</td>
<td>1</td>
<td>-1</td>
<td>F1</td>
</tr>
<tr>
<td>37</td>
<td>To receive updated and fact-based information.</td>
<td>2</td>
<td>3</td>
<td>3</td>
<td>F3</td>
</tr>
<tr>
<td>38</td>
<td>To receive information only about the tests that can be offered to me in my region.</td>
<td>-2</td>
<td>-4</td>
<td>-3</td>
<td>F2</td>
</tr>
<tr>
<td>39</td>
<td>To receive information about interpretation of test results, e.g. probability assessment with combined ultrasound and biochemical test.</td>
<td>2</td>
<td>3</td>
<td>1</td>
<td>F3</td>
</tr>
<tr>
<td>40</td>
<td>To receive the information in a way that makes me understand the implication (wording and knowledge level).</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>Consensus</td>
</tr>
<tr>
<td>41</td>
<td>To receive information about how long it takes to receive the test results.</td>
<td>0</td>
<td>-2</td>
<td>2</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>42</td>
<td>To receive information about anomalies or conditions e.g. Down syndrome only after a diagnosis has been confirmed.</td>
<td>-1</td>
<td>-2</td>
<td>1</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>43</td>
<td>To receive information about the life expectancy for individuals with Down syndrome.</td>
<td>-5</td>
<td>-3</td>
<td>-2</td>
<td>F3</td>
</tr>
<tr>
<td>44</td>
<td>To receive information that there might be a need for more follow-up tests to verify a diagnosis.</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>Consensus</td>
</tr>
<tr>
<td>45</td>
<td>To not receive information about prenatal diagnosis too early in the pregnancy.</td>
<td>-3</td>
<td>-4</td>
<td>-4</td>
<td>Consensus</td>
</tr>
<tr>
<td>46</td>
<td>To receive information from patient organizations or parents in similar situations.</td>
<td>-3</td>
<td>-3</td>
<td>-2</td>
<td>F3</td>
</tr>
<tr>
<td>47</td>
<td>To receive oral information in group.</td>
<td>-4</td>
<td>-4</td>
<td>-5</td>
<td>Consensus</td>
</tr>
<tr>
<td>48</td>
<td>To receive information about how adult life can be like for an individual with Down syndrome (medical care, living arrangements, occupation and support from society).</td>
<td>-4</td>
<td>-1</td>
<td>-2</td>
<td>F1, F2, F3</td>
</tr>
<tr>
<td>49</td>
<td>To receive information on what management and support is available during the course of the pregnancy if the fetus has an anomaly.</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>Consensus</td>
</tr>
<tr>
<td>50</td>
<td>To receive information on what causes Down syndrome.</td>
<td>-2</td>
<td>-2</td>
<td>-1</td>
<td>Consensus</td>
</tr>
</tbody>
</table>
Discussion

It is clear that decisions about PND are influenced by various factors, both individual and external. The levels of knowledge about DS among expectant parents and midwives seem to be generally low, and information provided to expectant parents is often insufficient. There is room for improvement in the information that the antenatal healthcare provides to expectant parents, regarding both the testing methods and the conditions screened for. To be able to make informed decisions regarding PND, adequate knowledge is essential. However, providing information and counselling to expectant parents is a complex task, as they have varying informational needs. Midwives should be given adequate education and resources to effectively address these needs.

In this section, different aspects of informed choice, information and counselling, knowledge of DS and decision-making will be discussed based on the results from the papers included in this thesis. Followed by some methodological considerations.

Informed choice

According to Marteau et al., a single-dimension measure (such as a measure of knowledge) does not reflect the multidimensionality of informed choice. However, to be able to choose whether or not to undergo PND, some knowledge about the condition screened for should be a prerequisite to enable an informed decision. In the papers included in this thesis, we did not assess the participants’ values. However, we found substantial knowledge gaps about DS among both expectant parents opting for screening or diagnostic testing and those declining PND. If we consider knowledge about the condition tested for as being important for making an informed choice about prenatal screening and diagnosis for CAs, it becomes necessary to ensure that pregnant women and their partners are well informed about DS and its consequences. This view is strengthened by the results from the included studies (Papers I, II, IV and V) and others indicating that expectant parents want to receive information about the conditions that can be detected with PND. The fact that the quality of life for an affected child and the expected burden on the family has an impact on decisions regarding PND (Paper IV) also highlights the need to provide information about the conditions tested for.
Facilitating informed choice is important, as knowledge and informed choice leads to improved well-being and reduces anxiety, decisional conflict and regret.\textsuperscript{121,131,134,136–140,143} Women who make more active decisions experience higher satisfaction with their choices, while perceived social pressure regarding their decisions may reduce choice satisfaction.\textsuperscript{169} Previous studies indicate that some women make the decision regarding PND as soon as they find out about their pregnancy, i.e. prior to pre-test counselling.\textsuperscript{36,170} It has been suggested that pre-test counselling might not change a decision that is already made, but the information could help rationalise their intuitive choice and could reduce decisional conflict.\textsuperscript{152,171} For these women and their partners, the aim of the counselling might not be to receive extensive knowledge about DS, but rather to get them to reflect on their decision-making and help them feel more at peace with their decisions.

Information and counselling to expectant parents

A majority of expectant parents, across all three groups in Papers I and II, felt they had received insufficient information about what it might mean to live with a child with DS. Many of those accepting CUB or invasive tests requested more information about DS. Despite this, when midwives were asked about the most common questions from expectant parents, inquiries concerning testing methods and risk assessments were the most common, while questions regarding symptoms of DS and the consequences of having a child with DS were rare. This can be perceived as somewhat contradictory to their stated need for more information. However, it has previously been discussed that satisfaction with information may not be related to knowledge but rather associated with having one’s expectations met.\textsuperscript{172} This could explain why women declining PND reported a higher satisfaction with the information they received compared to those undergoing invasive tests (Paper II). Women declining PND because TOP is not an option for them may not require extensive information about PND and DS. Women undergoing PND, on the other hand, are more likely to want more extensive information on which to base their decision,\textsuperscript{173} especially if they are unsure about what to do if they receive a DS diagnosis. Hence, the decision to accept PND might be driven by a need for more information concerning the pregnancy, while the choice to decline PND might be based more on feelings and values than a need for information. In addition, having already made a decision regarding PND before pre-test counselling might result in less need for information, regardless of whether one decides for or against PND.\textsuperscript{112} Being undecided when attending pre-test counselling may instead result in a need for more extensive information, e.g. about the individual probability of expecting a child with an anomaly.\textsuperscript{112}

A majority of midwives provided information to expectant parents about DS only upon request or in case of an increased probability. This tendency
might be a reflection of the fact that many midwives felt they lacked adequate knowledge to provide information about DS and its consequences. Additionally, the tight time schedule at the outpatient antenatal care units and the need to cover an extensive range of general pregnancy-related topics during a short visit could be another reason.\textsuperscript{27,115,174–177} Thus, midwives might prioritise providing information that they perceive parents most often request.

Ways to improve information and the knowledge of expectant parents could be written information, audio-visual information and booklet-based or web-based interactive decision aids.\textsuperscript{178–185} A randomised controlled trial with a web-based decision aid, including both audio-visual and written information about available tests for CAs (CUB, NIPT and invasive tests) as well as exercises for value clarification, showed good results with improved informed decision-making and decision-relevant knowledge concerning PND.\textsuperscript{186} Women randomised to the intervention group experienced less decisional conflict related to feeling uninformed.\textsuperscript{186} Similar results, with improved knowledge and informed decisions, as well as less decisional conflict after the use of computerised decision aid tools or educational video, have previously been reported from other randomised controlled trials.\textsuperscript{179,184,187} It has been argued that one advantage of decision aids is that they provide standardised information that is not affected by healthcare professionals’ own knowledge levels and opinions, and thus more likely to be accurate and non-directive.\textsuperscript{186} The International Society of Prenatal Diagnosis (ISPD) concluded in their 2023 Position Statement regarding the use of NIPT that pre- and post-test counselling is resource demanding and that research concerning alternative counselling methods should be explored.\textsuperscript{42} However, as shown in Paper V, and in other reports,\textsuperscript{13,25} many women prefer receiving verbal, individual information from healthcare professionals. In addition, based on the findings of Paper V regarding the varying informational needs of pregnant women, standardised information is less suitable. Thus, decision aids could be a good complement to individualised pre-test counselling provided by a trained professional, but they should not be seen as a replacement. Giving expectant parents access to a decision aid prior to individualised pre-test counselling with a healthcare professional, could reduce the time spent on providing basic test-related information and give more room for reflection and decision-making support during the counselling session.

As new test methods, such as NIPT, are introduced, offering easy-access testing early in pregnancy with no associated risks for the pregnancy, pre-test counselling becomes even more important. This is to ensure informed decisions and that the test is perceived as an offer rather than a routine.\textsuperscript{20,28,43,50,188,189} Concerns have been raised that it will be challenging to provide sufficient pre-test counselling to the large number of expectant parents who may want NIPT if it becomes easily accessible.\textsuperscript{68,189} SMER has concluded that, when NIPT is implemented, there is a need for educational efforts within the public antenatal care to enable proper pre-test counselling and that there is
a need for more genetic counsellors. Both SMER and SBU have emphasised the importance of including not only information on what the test can detect, but also information on the consequences for an affected child and what it is like to live with a child with a CA.

Healthcare professionals face a difficult task of providing sufficient information in a form and pace that a pregnant woman can effectively process, and get her to start reflecting on her decision to make it value-consistent. This requires communication and counselling skills. In addition, knowledge of what information is desired and what factors influence pregnant women’s decision-making can help guide healthcare professionals in this important task.

As shown in Paper V and another study, the informational needs of pregnant women vary. Some women prefer extensive information from the start, including different aspects of the conditions screened for and the decisions they may need to make regarding the pregnancy based on the test results. Others prefer a stepwise approach, with a focus on information and decision-making concerning tests as a first step. Additional reports have also discussed women’s use of a multi-step decision-making process, where information relevant to the immediate decision (e.g. screening) is preferred and prioritised over information pertaining to later decisions they may have to make (e.g. invasive tests or possible TOP). Notably, information about DS was still prioritised early in the process, even among some women who preferred the stepwise information process. Previous studies have indicated that women’s informational needs can be affected by parity, age and the probability of expecting a child with a fetal anomaly in a current or previous pregnancy. Despite this, Paper V did not find any significant differences in background characteristics, such as sociodemographic or coping styles, making it difficult to predict a woman’s informational needs. These results should, however, be interpreted with caution due to the small sample size tailored for the Q methodology. Women had a clear preference for individual information (Paper V). Given the varying informational needs, individualised pre-test counselling is preferable for discerning each woman’s needs and adapting the information accordingly. Both sufficient time and competence are needed to explore each individual woman’s counselling needs during pre-test counselling sessions. Moreover, healthcare professionals offering pre-test counselling to expectant parents should be aware that information about the conditions screened for is crucial for some women.

Knowledge of Down syndrome

Overall, knowledge levels regarding the medical, cognitive and social consequences of DS were varying, and in many respects low, both among expectant parents coming for CUB tests, invasive tests and declining PND for CAs as well as among midwives in the outpatient antenatal care. Various reports have
called attention to insufficient knowledge concerning DS testing among healthcare professionals in antenatal care.\textsuperscript{31–34,174,191} Thus, it may not be surprising that midwives’ knowledge about the condition being screened for is also insufficient (Paper III), as previously reported.\textsuperscript{33} Previous studies have also reported on insufficient knowledge of DS among expectant parents facing decisions about PND for CAs,\textsuperscript{27,117,125,126,128,129} which was confirmed by our results. Considering the varying and low levels of knowledge about DS among midwives who provide information to expectant parents, this is not surprising. In Sweden, it is common for partners to accompany the pregnant women to antenatal care visits, implying that they receive the same information about PND as the pregnant woman. Consequently, it is not surprising that partners had the same levels of knowledge as the pregnant women (Paper I), a finding in line with the results from a Danish study, where no differences in knowledge were found between women and partners.\textsuperscript{130}

It is worrying that midwives with the important task of providing pre-test counselling to expectant parents had varying, and in some areas low levels of knowledge about DS and that they did not feel competent to provide this information (Paper III). This is particularly concerning since many expectant parents, across all groups in Papers I and II, mentioned their midwife or doctor as the main source of information regarding PND, a finding that is consistent with other reports.\textsuperscript{10,12,13,25,26,31,192,193} However, considering midwives’ feelings of insufficient knowledge, and the fact that few of them reported giving information about DS as a routine to all expectant parents (Paper III), it is not surprising that midwives were not the main source of information regarding DS for many expectant parents. The Internet and friends seem to play a more important role as sources of information regarding DS, which is not surprising considering previous reports on pregnant women’s Internet use.\textsuperscript{194} That healthcare professionals may feel less comfortable in providing information related to conditions tested for, compared to test characteristics, and less often discuss such issues with expectant parents has previously been reported.\textsuperscript{175}

It is remarkable that only 3\% of midwives in Paper III had received postgraduate education related to PND. Guidelines from The National Board of Health and Welfare states that it is the responsibility of the healthcare provider to ensure that healthcare professionals receive adequate education and have sufficient competence to provide information about and offer PND.\textsuperscript{21} A vast majority of midwives requested more education in all areas regarding PND and DS (Paper III), similar to other reports where healthcare professionals valued continuing professional training regarding PND and conditions screened for.\textsuperscript{32,34–36,191} These findings indicate that their lack of knowledge is not due to lack of interest, but rather due to limited resources and lack of adequate education. The results from Paper III indicate that midwives’ knowledge was not always up-to-date as, e.g. one-third of them underestimated the mean survival age for individuals with DS, stating it was 30 years, which was true some decades ago. Previous studies have also indicated insufficient knowledge of
PND and DS among healthcare professionals counselling women about PND, as well as low self-perceived knowledge. It is worth noting that the finding in Paper III, indicating that the length of time working as a midwife was not associated with knowledge, is congruent with later research. Moreover, the finding that midwives’ self-perceived knowledge did not always correspond to their actual knowledge is interesting. Similar findings have been reported previously.

To ensure that midwives have the ability and sufficient knowledge to facilitate informed decision-making for expectant parents by providing well-balanced and good-quality information, it is crucial that they receive education and support needed to cope with this important task. There is a need for educational efforts and increased support for healthcare professionals in antenatal care, especially in relation to the implementation of NIPT. In recent years, after Papers I-III were published, efforts have been made to educate midwives in antenatal care, with a focus on pre-test counselling, in several regions in Sweden (e.g. Uppsala where participants for Papers I-III were recruited). Studies from other countries have shown a positive impact of continuing professional training with higher knowledge levels concerning PND among healthcare professionals who had participated in professional training. Thus, if Study III was repeated today, the results would hopefully be improved as a result of the in-service training aimed at improving midwives’ knowledge and ability to provide high-quality pre-test counselling.

Decision-making regarding prenatal diagnosis

Knowledge about DS, on the levels as shown in our studies, does not seem to affect expectant parents’ decision to accept or decline PND for DS. It is, therefore, evident that other factors are involved in the decision-making process. Our interview study provides a deeper understanding of the complex decision-making process, which is influenced by both individual and external factors on different levels.

Pregnant women put much emphasis on test-related risks, and for many this was a deal-breaker as they were not willing to risk a procedure-related miscarriage (Papers II and IV). Other test-related factors, such as timing, easiness and test accuracy, also influenced the decision-making processes for women (Paper IV). The impact of these test characteristics on women’s decision-making regarding PND has previously been reported. In light of these findings, it is not surprising that NIPT is the test of preference for many women, as it is easy to perform, can be done early in pregnancy and has a higher detection rate than CUB, but without the risks associated with invasive PND. However, since neither CUB nor NIPT are associated with a procedure-related risk of miscarriage, it is possible that expectant parents
might choose to undergo such tests without fully considering the consequences it could entail related to their decision-making.

One-fourth of the expectant parents who came for a CUB test (Paper I) had not yet decided if they wanted to undergo invasive PND for diagnostic purposes if given a high CUB probability. Additionally, almost half of these expectant parents had not yet decided what they would do if DS was diagnosed. This, in combination with statements such as “we did the test because you should if older than 35”, raises concerns about whether expectant parents view the CUB test as a routine procedure rather than an option. Several studies have highlighted the risk of routinisation regarding both CUB and NIPT, and a systematic review concluded that there is a risk that expectant parents and professionals regard screening tests as routine procedures that do not require active decision-making. Our findings also indicate that as many as one-fifth of women undergoing invasive PND may not have finished their decision-making process either, as they had not yet decided what they would do if DS was diagnosed (Paper II). According to a review investigating decision-making following a prenatal diagnosis of DS, the decision to terminate the pregnancy varied depending on whether participants were expectant parents recruited from the general population (23-33% would terminate), pregnant women at increased risk of having a child with DS (46-86% would terminate), or women who received a diagnosis of DS during the prenatal period (89-97% terminated). These findings suggest that women who undergo an invasive test have gone through a more meticulous decision-making process compared to those who undergo a screening test. Whereas invasive tests are diagnostic and carry a procedure-related risk of miscarriage, screening tests with no risks can be perceived as routine and might be decided on more casually. Furthermore, more women undergoing invasive tests could consider having a TOP following a DS diagnosis compared to those declining PND for CAs (Paper II). Women who would consider a TOP in case of an anomaly may be more inclined to undergo invasive testing, despite the risk of having a procedure-related miscarriage.

Besides the risk of routinisation, another concern when risk-free screening tests become more easily accessible is that women may lose an ‘excuse’ to decline testing. It has been suggested that the risk of miscarriage may be used as a justification, either to themselves, a partner or to healthcare professionals, to decline the offer of PND, when, in fact they just do not want to know or do not want to deal with the results. Depending on which tests are offered and how they are offered, pregnant women may feel a perceived social pressure from society in general either to undergo testing (and have a TOP if a CA is diagnosed) or to refrain from testing and TOP. Previous research has indicated that women who have a religion that condemns TOP, or that live in a country where TOP is illegal are less likely to undergo PND. The self-perceived probability (or ‘risk’ as described by most women in Paper IV) of expecting a fetus with an anomaly seems to play a part in the
decision-making process for some women, as indicated in Papers I, II and IV. Age seems to be one of the most important factors when women assess their own probability (Paper IV). Advanced maternal age can motivate testing, while younger women may not feel the need for it (Papers I-II). It is evident that many pregnant women are aware that with an increased age comes an increased probability of CAs. One might suspect that the fact that PND for CAs has traditionally been offered primarily to older women, and in some regions in Sweden still is, probably have highlighted the influence of age. To pregnant women, this can emphasise the increased probability when pregnant at a more advanced age (possibly making them feel obliged to have PND), while reassuring younger women that their probability is relatively low (possibly making them feel that they should not choose to undergo PND).

The tests that are offered, and how healthcare professionals present both the tests as well as a woman’s individual probability of expecting a fetus with a CA can influence decisions concerning PND (Paper IV), as previously reported. Through their counselling style, wording and content, healthcare professionals may unintentionally influence pregnant women’s decisions. To be offered a test by a ‘trusted’ healthcare professional may result in a perceived need to undergo testing. Moreover, the fact that the public antenatal care has allocated time and money to actively offer PND may signal to women that these tests are offered for a reason, and be perceived as a recommendation or part of routine care. In contrast, to not be offered a test could hinder a woman’s access to it, even if she may have wanted it (Paper IV). For some women, not being offered a test is a reason to refrain from testing, either because they perceive it as a signal from healthcare that there is no need for testing or because having a test privately in another region would cost them both time and money (Paper IV). The cost of a test and its effect on decisions about PND have previously been highlighted, raising concerns that having to pay out-of-pocket for PND may be a barrier to testing for some individuals. Offering PND under unequal terms, with different age-limits and costs is problematic, as guidelines from the National Board of Health and Welfare stipulate that PND should be offered to all women on equal terms. This principle was recently emphasised by a Swedish expert group that reviewed the offer of PND in Sweden. However, other reports suggest that while the final decision outcome may not be influenced by having to pay for a test, it could affect the decision-making process by making women aware that the test is optional rather than a routine procedure. Nonetheless, if we are to achieve informed choices, women’s decisions to undergo or abstain from PND should be based on their own personal values and needs, rather than the organisation of the healthcare system and the tests available to them.

In Paper II, we found that a considerable number of women who declined PND felt they could manage to care for a child with DS. Often, DS was not regarded a severe enough condition to justify TOP, congruent with previous
reports. Moreover, many women who declined PND did so because they did not regard TOP as an option, also reported previously. In contrast, women who opted for CUB or invasive testing did not necessarily associate PND with TOP if DS was diagnosed. Rather, they saw it as a way to gather information, to be reassured that the fetus was healthy and to ease their worries. These results resemble previous observations and were confirmed by the findings from Paper IV. Some women that would not consider TOP still saw a value in undergoing PND, as knowing about an anomaly in advance allowed them to prepare for the birth of an affected child (Paper IV). In addition, some women in Paper IV regarded the CUB test not primarily as a probability assessment for CAs, but rather as an extra opportunity to have an ultrasound and see the baby. Undergoing an ultrasound scan can make the pregnancy feel more real, and seeing the fetus is a strong emotional experience that can facilitate expectant parents’ bonding with their unborn child. However, as shown in Paper IV, worry can also be a reason to abstain from PND, as having PND may cause extra anxiety or worry.

The expected quality of life for a child with a condition influences women’s decisions regarding PND and TOP (Paper IV). Additionally, the perceived impact that an affected child would have on the parents and siblings is also taken into account when making decisions about PND (Paper IV). Some women are more inclined to test for what they perceive as more serious conditions, and the severity of the condition may also affect decisions regarding TOP. In Paper IV, what was considered a severe condition motivating testing varied, probably due to previous experiences and attitudes towards anomalies. Women who declined PND in Paper II more often had personal experience of someone with DS in their vicinity compared to women who accepted PND (CUB or invasive tests). Perhaps personal experience could result in a more positive attitude towards DS, making women more inclined to abstain from PND. For those knowing someone with DS, there might also be an element of guilt or perceived disapproval from others involved when making decisions about PND. However, previous reports suggest that having a sibling with DS can, for some women, result in a more negative view of DS due to a perceived negative impact on them and their family. Women who have a child with DS or know a child with DS may be more inclined to undergo testing. The perception of how a family is affected may differ depending on whether someone with DS is among friends or within the immediate family. In this context, the findings in Paper II, that some women chose to have invasive tests because of DS or a sick child in the family, while others who knew someone with DS declined such tests, may not appear as contradictory as it might first seem. In Paper IV, it was clear that personal experience of anomalies in the family influenced women’s perceptions of anomalies and disability, albeit in different ways. Knowing the physical and emotional burden it might bring to the family made
some women more inclined to have PND. Conversely, for others, such experience made them less inclined to have PND, as they felt they could handle it and an affected child would be welcomed and bring them much love. Moreover, opting for PND for DS does not automatically mean the intent is to terminate an affected pregnancy, but it can be a way to prepare.

When comparing the reasons why women and partners choose a CUB test, we also found that significantly more women than partners chose the test because of worry (Paper I). Previous studies have also indicated that women and their partners have different reasons for undergoing PND. Women are often influenced by their partners in their decision-making process regarding PND, and often strive for a consensus decision. The need to involve partners in the decision-making process varied in Papers IV and V. For those who wish to discuss their decision with their partner, there is a need for time between receiving information and making a decision. Healthcare professionals who provide information to expectant parents need to be aware that the extent to which a pregnant woman wishes to involve her partner in the decision-making process varies. They need to be flexible in involving the partner when information about PND is given, while keeping in mind that decisions concerning PND are ultimately the woman’s choice, although both expectant parents are affected by the decision.

Methodological considerations

This thesis is based on five papers that have included pregnant women and partners, as well as midwives working in antenatal care. In addition, three different methods have been used: quantitative, qualitative and Q methodology. By exploring the perspectives of women, partners and midwives we gain a better understanding of the complex information and decision-making process. The use of different methods provides a broader and deeper understanding. A general weakness of all the included papers is that, for practical reasons, only Swedish speaking individuals were included. This may affect the generalisability of the findings, as not all pregnant women and partners in Sweden have a sufficient command of the Swedish language. By not including non-Swedish speakers in the studies, it is possible that we overlooked their perspectives.

Papers I and II

The study design and recruitment of participants were similar for both Paper I and II. The questionnaires were designed specifically for the study and were not pilot tested or validated. They were, however, developed by a research team, including different backgrounds (fetal medicine specialists, a genetic counsellor and a paediatrician specialised in CAs), each contributing with different perspectives on the subject. Knowledge questions were of multiple-
choice format to facilitate comparative analyses. However, this might have failed to capture the full range of contextual factors involved. To encourage open and honest answers, the questionnaires were completely anonymous. Due to the anonymous design, however, non-participants could not be followed up, which is a weakness. Nonetheless, we have no reason to believe that participant recruitment was systematically skewed. Participants filled in the questionnaires during their visits to undergo the CUB test, invasive test or second-trimester ultrasound. This is a strength, as it reflects the opinions held by the participants when it was of immediate interest. However, since those who had undergone CUB or a second-trimester ultrasound had already received their results, their views on invasive tests and TOP might have been influenced by the results received. Women and partners, included as a couple for Paper I, were instructed to fill in the questionnaires independently without discussing them with each other. However, we cannot be sure that they did this, which may have affected the results of comparative analyses between women and partners.

A power calculation was conducted concerning the knowledge questions. In order to achieve a power of 80% at a significance level of 0.05, a sample of 94 participants would be needed in each group to detect a difference of 20 percentage units. This calculation assumed that 50% of the control group (partners for Paper I, and those declining PND for Paper II) would answer the knowledge questions correctly. However, since many women chose to participate in the CUB screening programme, we had difficulties in recruiting participants coming for invasive tests and women abstaining from PND for CAs for Paper II. Thus, in order to avoid a too long study period, only 76 participants in the invasive test group and 65 in the declining group were included in Paper II, which is a limitation. With this number of participants, a difference of 23 percentage units could be detected.

**Paper III**

The strengths and weaknesses of the questionnaire design, with multiple choice response alternatives and a study-specific questionnaire, were the same as for Papers I and II. Moreover, as the knowledge questions were the same for Paper III, no new power calculation was needed. Since the aim was to explore the perspectives of midwives in the same county as expectant parents of Papers I and II, the number of participants was restricted to those working in the county, which was insufficient for internal comparisons according to the power calculation. However, a majority of the midwives working in the county participated, which is a strength. Thus, these findings are representative of the knowledge and opinions of midwives from one of Sweden’s 21 counties, possibly restricting the generalisability somewhat. Midwives’ undergraduate and postgraduate education is, however, similar across the country, so it is not unlikely that the results would have been similar in other counties.
Paper IV
The qualitative approach complements earlier quantitative research by providing a deeper understanding of various aspects that influence women’s decision-making. Most of the women were approached and interviewed in early pregnancy, before they had met with a midwife and received information regarding PND and conditions screened for. This is a strength, as information from a healthcare professional prior to the interview could have influenced their views on the subject. However, it is unclear if parous women were influenced by discussions about PND and CAs that they may have had in previous pregnancies. The diverse participant group, including different parity, age and educational levels, increases the transferability of the findings. Another strength is that the analysis and interpretation were performed by a research team having different professional backgrounds relevant to the research question, giving complementary perspectives to the interpretation of the results. As with any qualitative research, no claims of generalisability can be made, but the findings are in line with previous qualitative and quantitative research and give us a picture of the factors influencing the decision-making process regarding PND.

Paper V
By including women with diverse backgrounds, a good representation of pregnant women in the Swedish healthcare system was achieved. Thus, similar viewpoints are likely to be found among other pregnant women facing similar circumstances. The aim of Q methodology is not to gather a large participant group, but rather to include participants with diverse backgrounds and views on the subject and stop recruitment once saturation has been reached. Therefore, comparative analyses between the factors must be interpreted with caution, as minor differences may go undetected due to small sample sizes. Bigger differences should still emerge. Q methodology often includes qualitative data from interviews, but this was not possible because of the Covid-19 pandemic. Thus, qualitative insights for factor interpretation were derived from written comments. This presents a potential weakness, as in-person interviews could have provided a deeper understanding of the viewpoints.
Conclusions

- Expectant parents making decisions about accepting or declining prenatal diagnosis (PND) for Down syndrome (DS) have varying, and in several respects, low levels of knowledge about DS and its consequences (Papers I-II).

- Knowledge of DS, at the levels found in Papers I and II, is not a major factor when women decide to accept or decline PND for DS. The decision-making process regarding PND is influenced by individual factors, such as attitudes towards anomalies, worry and need for reassurance, self-perceived risk, and by external factors such as test characteristics and influence from others (Papers I, II, IV).

- Prenatal tests appear to be perceived as routine procedures by many expectant parents undergoing combined ultrasound and biochemistry (CUB) tests and by some undergoing invasive tests. In many cases, the decision-making process seems to be incomplete, as expectant parents remain undecided about undergoing invasive tests following a high CUB probability, and regarding the decision to continue or terminate the pregnancy in case of a DS diagnosis (Papers I-II).

- Providing information about PND and conditions tested for is complex. Some women prefer a step-by-step process, while others want extensive information, including details about conditions tested for from the start. Pregnant women’s varying informational needs make individual pre-test counselling important, so that the information can be individualised to meet the different needs (Paper V).

- Information about DS is mostly provided by midwives when requested or when there is an increased probability of a DS pregnancy. Expectant parents report that much of their information about DS comes from sources other than healthcare professionals (Papers I-III).

- Midwives providing information about PND to expectant parents in the outpatient antenatal care have varying, and in some areas low levels of knowledge about DS and its consequences (Paper III).

- A large majority of midwives reported insufficient or no education regarding different prenatal tests and DS. They expressed a desire for more in-service training about second-trimester ultrasound, invasive testing, CUB and DS (Paper III).
Providing information about PND and conditions tested for is complex as women’s informational needs differ. Our results can provide guidance for healthcare professionals working in antenatal care concerning the information expectant parents wish to receive, and provide a better understanding of factors that influence the decision-making process regarding PND. With a better knowledge of these varying informational needs and the factors that influence expectant parent’s decisions about PND, a better understanding of the importance of individualised pre-test counselling, with healthcare professionals trained for the task, is gained. Counselling should be a two-way communication aimed at discerning each individual pregnant woman’s informational needs. The aim is to provide sufficient individualised information and evoke reflection about the decision, to facilitate informed decisions. In this process, healthcare professionals need to be aware that their approach and attitudes can influence the decisions of expectant parents. Decisions may be affected by how tests and conditions are presented and offered. Moreover, pregnant women’s self-perceived probability can be affected by how they perceive that their midwife has made an assessment of their individual need for having PND. This highlights the need for non-directive counselling with emphasis on offering PND as an option and not as routine tests.

It is evident that information about test characteristics (e.g. test-related risks) is crucial to many expectant parents, and such information is often provided during pre-test counselling. Additionally, women’s decisions about PND are also influenced by their perception of how a condition might affect the quality of life for an affected individual and his or her family. Therefore, knowledge about the condition being tested for is also essential when making informed choices about PND for CAs. Hence, some information about the conditions being tested for should be an integral part of the information provided prior to decisions about PND are made. However, not all pregnant women wish to receive such information from the start, but prefer to wait until screening indicates an increased probability. To explore the needs of each expectant woman during pre-test counselling sessions is therefore crucial. This research highlights the need for improved information to expectant parents. Midwives in the outpatient antenatal care are important providers of this information and counselling. In order for midwives to feel confident about this important task, they need more education about PND and DS and adequate
resources. Since Papers I-III were published, a few counties have introduced supplementary in-service training for midwives working in outpatient antenatal care. This is an important step towards ensuring that expectant parents receive the information and counselling they need to make informed decisions.

Several reports have pointed out the influence that the expected quality of life and impact on family life of a condition can have on decisions about PND. Despite this, research regarding informed choice and knowledge often focus on knowledge about the different prenatal tests, with a limited focus on knowledge about various aspects of the conditions tested for. Further research on information and knowledge about conditions being tested for and its effect on informed choice could provide more guidance for healthcare professionals involved in pre-test counselling. It would also be interesting to further explore the effect of coping styles on informational needs related to PND in a study with a larger sample size.

It is also worth noting that the current arrangement with an unequal offer of PND (with different age-limits and costs) across different regions of Sweden is problematic. Not only because it does not adhere to the standards of equal access to healthcare across the country, but also because it may hinder informed choices as some women may not have a chance to make value-consistent choices if they are not offered PND. In light of this, the organisation of PND services needs to be discussed further. In addition, the possibility of providing a first-trimester ultrasound as an alternative to CUB for those who do not wish to receive a probability assessment, but still wish to have an ultrasound, should be explored in those counties that have not yet implemented such alternatives.
Bakgrund
Fosterdiagnostik för kromosomavvikelser erbjuds som en del av mödravårds- och en ca 70 % av gravida kvinnor genomgår screening med kombinerat ultraljud och biokemi (KUB-test). Enligt lagen om genetisk integritet ska alla gravida kvinnor erbjudas allmän information om fosterdiagnostik och enligt Socialstyrelsens riktlinjer ska denna information erbjudas vid inskrivningssamtalen på mödravårdscentralen. Informationen om fosterdiagnostik för kromosomavvikelser fokuserar ofta på sannolikheten att föda ett barn med en kromosomavvikelse, framförallt Downs syndrom (DS), och riskerna associerade med de invasiva diagnostiska testerna (fostervatten- och moderkaksprov). Ofta ges ingen eller sparsam information om DS och konsekvenserna som detta tillstånd kan ha för barnet och familjen. Enligt Statens Medicinsk-Etiska Råd bör informationen omfatta även medicinska och psykosociala aspekter av DS.


Tidigare forskning har mestadels fokuserat på information om testerna, men några studier har visat att gravida kvinnor har varierande eller dåliga kunskaper om DS och dess konsekvenser. Även om det i slutänden är kvinnan som fattar beslut om fosterdiagnostik strävar många efter att fatta ett gemensamt beslut som par. Trots detta inkluderas ofta inte de
gravidas partners i studier kring information om, och informerat val gällande fosterdiagnostik.

Barnmorskor och läkare är viktiga källor till information om fosterdiagnostik för gravida kvinnor. Samtidigt har studier visat att sjukvårdspersonal som informerar blivande föräldrar om fosterdiagnostik har kunskapsbrister gällande fosterdiagnostik för DS, att de upplever sig ha otillräcklig kunskap och är positiva till mer fortbildning inom området.

Inför beslut om fosterdiagnostik lägger gravida vikt vid information om risker med de olika fosterdiagnostiska testerna. En del önskar även information om DS och sannolikheten att vänta ett barn med DS tidigt i graviditeten. Tidigare rapporter indikerar att beslut gällande fosterdiagnostik kan påverkas av den förväntade livskvaliteten för ett barn med en avvikelse, samt påverkan på föräldrarnas och familjens liv. Informationsbehovet tycks också påverkas av en kvinnas sannolikhet att vänta ett barn med en avvikelse i en tidigare eller pågående graviditet.

Eftersom tidigare studier indikerar otillräcklig information och kunskap hos blivande föräldrar som fattar beslut om fosterdiagnostik finns det ett behov att närmare undersöka blivande föräldrars och vårdpersonals kunskapsnivåer om DS och informationen som ges. Det behövs en förджупning och mer fokus på föräldrarnas perspektiv beträffande vilken information de önskar och hur de vill få den för att kunna göra informerade val. Det är nödvändigt för att få kunskap om hur vägledningen kring fosterdiagnostik bör organiseras och vilken information den ska tillhandahålla.

Syfte

Syftet med de olika delarbetena i denna avhandling har varit följande:

I. Att undersöka blivande föräldrars kunskap om DS och vilken information de fått innan de gör KUB-test.

II. Att undersöka orsaker till att genomgå eller avstå fosterdiagnostik, samt om kunskap om DS skiljer sig mellan de som genomgår eller avstår fosterdiagnostik.

III. Att undersöka barnmorskors kunskap om DS och fosterdiagnostik, samt vilken information de ger till blivande föräldrar.

IV. Att undersöka vad som påverkar beslutet att genomgå eller avstå fosterdiagnostik.

V. Att undersöka vad som är viktigt för gravida kvinnor när de får information om fosterdiagnostik gällande innehåll och sätt att få information.

Metod

Studie I-III var enkätstudier med deskriptiva och jämförande analyser. I studie I och II tillfrågades blivande föräldrar som kom till den fostermedicinska mottagningen i Uppsala för KUB-test, invasiv provtagning eller rutinultraljud (om de valt att avstå kromsominriktad fosterdiagnostik) om deltagande och fyllda
i enkätarna direkt på plats. I studie III fyllde barnmorskorna i en enkät i sam-
band med en fortbildningsdag. Alla enkäter var anonyma. Jämförande ana-
lyser gjordes bl.a. med avseende på kunskapsnivåer om DS mellan kvinnor
och partners i studie I, mellan de som gjorde invasiv provtagning och de som
avstod kromosominriktad fosterdiagnostik i studie II och mellan barnmorskor
som jobbat längre eller kortare i studie III.

Studie IV var en kvalitativ studie baserad på individuella telefonintervjuer
med gravida kvinnor som tillfrågades om vad de såg som viktigt vid information om
fosterdiagnostik och prioritera mellan dessa från minst viktigt till mest viktigt.
Påståendena togs fram baserat på aktuell litteratur, intervjuerna från studie IV,
kommunikation med mammor med barn med DS och genom diskussioner
inom forskargruppen. Med Q-faktoranalys fick vi fram grupper med gravida
som sorterat påståendena likartat och därmed delar samma subjektiva syn på
ämnet. De gravida fick även lämna skriftliga kommentarer och fylla i 2 enkä-
ter, en studiespecifik enkät med sociodemografiska bakgrundsfrågor och en
validerad enkät för att bedöma copingstilar (Threatening Medical Situations
Inventory (TMSI)). Faktorerna (grupperna med gravida som delade samma
syn på ämnet) tolkades med hjälp av resultaten från Q-faktoranalysen, kvali-
tativa kommentarer från de gravida om sorteringsprocessen samt bakgrund-
data. Jämförelseanalyser gjordes mellan de tre grupperna som faktoranalysen
resulterade i med avseende på bakgrundskarakteristika och copingstilar.

Resultat och slutsatser

I. Blivande föräldrar som genomgick KUB-test hade varierande till låga
kunskapsnivåer om DS, med få skillnader mellan gravida och partners.
En del blivande föräldrar verkar se KUB-test som en rutinundersökning.

II. Gravida som genomgick invasiv fosterdiagnostik och gravida som av-
stod kromosominriktad fosterdiagnostik hade varierande till låga kun-
skaper om DS. Inga signifikanta skillnader i kunskap hittades mellan
grupperna, således verkar inte kunskap om DS på de nivåer vi fann på-
verka beslutet att genomgå eller avstå fosterdiagnostik. Fler som avstod
fosterdiagnostik kände någon med DS.

III. Majoriteten av barnmorskorna upplevde att de fått otillräcklig utbild-
ning om fosterdiagnostiska tester och få tyckte att de hade tillräcklig
kunskap för att ge information om DS. Nästan alla önskade mer fort-
bildning både gällande tester och DS. De faktiska kunskapsnivåerna
gällande medicinska, kognitiva och sociala konsekvenser av DS var va-
rierande och i vissa avseenden låga.

IV. Beslut om fosterdiagnostik är multidimensionella och påverkas av egna
faktorer (såsom attityd till avvikelser, oro och behov av bekräftelse och

63
självupplevd risk) samt externa faktorer (såsom testens egenskaper och påverkan från andra). Förväntad livskvalitet för en individ med en avvikelse och hur det påverkar övriga familjen påverkar beslutet för en del gravida. Vårdpersonal som ger information till blivande föräldrar kan påverka gravidas beslut om fosterdiagnostik genom sin attityd och sättet de presenterar såväl testerna som kvinnans individuella sannolikhet att vänta ett barn med en kromosomavvikelse.

V. Gravida kvinnors informationsbehov varierar. Vi fann tre olika synsätt som återspeglar olika informationsbehov, där vissa vill ha stegvis information och beslutsprocess medan andra vill ha mycket information tidigt och även ha information om tillstånden man testar för och hur de kan påverka livet för barnet och familjen. Resultaten talar för att individuell rådgivning anpassad efter de blivande föräldrarnas behov är att föredra för att möjliggöra informerade val. Information i grupp var generellt lågt rankat.

Sammantaget visar dessa studier att blivande föräldrar inte alltid har tillräcklig kunskap om DS när de fattar beslut om fosterdiagnostik och att informationen om fosterdiagnostik och kromosomavvikelser behöver förbättras. Gravida kvinnors beslut om fosterdiagnostik påverkas av både individuella och yttre faktorer och de har olika informationsbehov. Dessa behov tillgodoses bäst vid enskilda samtal med utbildad vårdpersonal som kan ge information och rådgivning.
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Appendix

Interview guide (Paper IV)

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<th>THEME</th>
<th>QUESTIONS</th>
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| 1) Information that is relevant for expectant parents being able to make informed choices about prenatal diagnosis. | - What are your thoughts about prenatal diagnosis?  
- Describe the grounds on which you make decisions about prenatal diagnosis.  
- What information do you need in order to make a decision?  
- What information about the conditions that are tested for, do you think is important/relevant to receive prior to prenatal testing.  
- When would you prefer to receive information about prenatal diagnosis and the conditions that can be identified – prior to testing or when one receives a result showing high risk? |
| 2) How does it feel to make decisions about prenatal diagnosis and what factors play a role in the decision? | - How difficult do you think it is to make a decision about declining or undergoing prenatal diagnosis?  
- What factors play a role in the decision?  
  - If the test is invasive or not?  
  - How early in the pregnancy the test can be taken?  
  - How long it takes to receive an answer?  
  - What can be detected?  
  - The test’s accuracy/inaccuracy, false positive/false negative results? |
| 3) Professional demeanour in conjunction with receiving information about prenatal diagnosis | - What do you think is important in the demeanour of healthcare professionals when you receive information about prenatal diagnosis? |
| 4) Expectant parents’ attitudes and reflections about Down syndrome. | - How do you regard having a child with a disorder?  
- What are your thoughts about Down syndrome?  
- How do you think having a child with Down syndrome would affect you?  
- How do you think it would affect your relationship, your family, your career etc.? |
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