

# **Prenatal diagnostics – 23 theses**

## Preliminary remarks

These 23 theses represent the gist of our discussions. The complexity of the topic did preclude a complete representation of the discussion and we also would not want to suggest that we raised or even pondered all relevant questions. The theses represent points of debate that seemed particularly important to us and through which we would like to stimulate a continued discussion beyond our group. Individual themes reappear in distinct theses, though in part from different perspectives, with a different emphasis or different target, at the same time showing links and associations.

All theses are the result of a consensus process because it was important to us to represent the plurality of the group and to withstand any tensions. This is mirrored in the dichotomy of some theses and their orientation, but it also shows a consensus in the group. While the theses thus do not express the positions of all participants, they nonetheless represent positions that were at least accepted as relevant to the discussion. We do not claim to have reached comprehensiveness. Rather, we are aware that our discussion has not been concluded but would like to provide an insight into the state of our discussions now to prompt a continued societal discourse involving all disciplines. This will lead to further themes being added, showing new connections while some theses will stimulate critique and contradiction.

## **1. Provide equal access to individual tests for all pregnant women**

All prospective parents share the desire for a healthy child. This concern makes them a target for those offering additional prenatal testing methods not covered by general health insurance.

In fact, relevant tests such as early or late ultrasonography or laboratory tests are often withheld from pregnant women without medical indication and with reference to the German guidelines for pregnancy care (*Mutterschaftsrichtlinien*), and instead offered as an additional service at their own expense.

Experience has shown that accurate anamnesis would often justify examinations as part of the national health service coverage. It is, however, extremely difficult for pregnant women to verify the need for such tests. In addition, medical assistants are frequently trained to offer tests tailored to patients at patient expense. The medical profession has thus created false incentives within the health care system itself. Information on available tests is tailored to parents' desires and concerns, which can create a misleading perception of certainty and control. Socially disadvantaged families are easily excluded from the available offers.

It is, in part, difficult for patients to decide whether and which kind of additional testing would indeed be useful. This appears to be the case in particular if physicians that women

trust in and rely on more or less consciously pursue their own financial interests. It is also known that, in general, patients often readily accept to pay high costs for medical services suggested to them [1].

Legislators would be in a position to support pregnant women with easily accessible and clear information. It may also be necessary to reconsider professional self-regulation of physicians to protect pregnant women from the effects of increasing commercialisation.

Pregnant women should be offered neutral counselling and receive only tests tailored to their specific situation. Every pregnant woman should have a wide-ranging offer of high-quality and reliable testing methods at her disposal as part of general health insurance and enabling prospective parents independently from their financial situation or their educational background to make considered and informed decisions for or against relevant tests. Moreover, concerns about malformations of the unborn should not be instrumentalised and misused as a basis for offering additional tests at patient expense and increasing commercialisation [2]. Instead of nourishing concerns, the focus should be on equal access to high quality health services and the avoidance of a socially stratified health care system.

## **2. Improve access to information about both potential and limitations of available testing methods**

If tests during routine pregnancy care indicate risks to the fetus, the pregnant woman or a high-risk pregnancy, it is the responsibility of physicians to arrange for additional genetic counselling and/or ultrasonography or laboratory tests.

Familial genetic dispositions can increase the risks for prospective mothers. Pregnancies of women above the age of 35 have an increased risk for trisomy 21, 18 and 13. In Germany, all tests that are suitable for establishing genetic risks are subject to the German law on genetic testing (*Gendiagnostikgesetz*) and require genetic counselling. These methods include first trimester screening, the triple or quadruple test, obstetric ultrasonography, non-invasive molecular genetic prenatal testing (NIPTs) and in particular invasive diagnostic methods such as amniocentesis or placental tissue sampling.

Basic or level 1 ultrasound screens are, however, not subject to the law since these are not aimed at the detection of genetic disease.

Non-invasive methods do not provide a diagnosis as a result, but probabilities concerning the occurrence of trisomy 21, 18 or 13. A negative test result does not prove that the fetus is healthy, while a positive result does not imply that the fetus is affected. A reliable diagnosis can only be established via invasive methods such as amniocentesis or chorionic villus sampling.

Consequently, pregnant women have to weigh the risk of miscarriage related to an invasive test with the remaining risk of being affected by trisomy 21. This situation is demanding on many women while only few are aware of the existence of 'false positives' and how these can contribute to uncertainty. The probability of discovering trisomy 21 lies within a range of 50% (25% false positive rate at age 35) and 99% (NIPT false positive rate 0,1%) depending on testing methods [1]. Pregnant women are often not being informed sufficiently about the relevance of detection rates and false positive rates. On the contrary, leaflets and information material in circulation tend to refer to terms such as "security" and "(secure) knowledge", while drawing on prospective parents' desire for reassurance. Even the result of NIPT, however, does require subsequent invasive testing in case of positive findings to substantiate the result. Invasive testing is also performed to exclude chromosomal changes in the case of heart defects or other malformations.

The multitude of methods available means that professionals increasingly require specific knowledge and training in risk communication to support the informed consent of pregnant women (cf. thesis 4). Yet, PND of any kind is unable to provide a 'guarantee' for a healthy child. Pregnant women are encouraged to seek information about PND before use. It must be highlighted, however, that information available online is not always reliable (cf. theses 18-20). To support this process and unpressured decision-making, pregnant women are advised to make use of psycho-social counselling provided by specialised counselling providers (cf. thesis 11).

### **3. Improve the quality of ultrasound scans**

In 2014, 9,665 gynaecological practices in Germany [1] each served around 60 pregnant women per year [2]. This wide distribution does not support reaching sufficient expertise of ultrasound scanning for rare diseases. Consequently, it may appear unjustifiable to equip all practices with cost-intensive and up-to-date diagnostic devices rather than advancing the development of specialised service practices and PND centres.

Basic ultrasound scans are covered by general health insurance since 1980. Since 2013, an additional scan to evaluate the organ development of the fetus is available following patient counselling [3].

Only sufficient evidence that a woman is at risk allows for secondary tests to be provided by specialist medical professionals with high-quality testing methods at their disposal and to be covered by general health insurance. Initial evidence of this kind, however, often remains undetected, with a persistently low detection rate for prenatal malformations (for live births) at 25-33% [4].

It is known that increasing expertise of professionals and higher-quality scans lead to higher detection rates [5], as is that the quality of tests remains poor without quality control [6]. As a consequence, pregnant women should all have access to the significantly

higher quality tests of level 2 [7]. In addition, pregnant women should be offered independent, open-ended counselling, which outlines advantages and disadvantages of screening and thereby facilitates informed decision-making.

The differing qualification of test providers is difficult to assess for users and requires an advanced level understanding of the issues involved. Quality controls of ultrasound screening level 1 as well as level 2 (to evaluate preliminary results) are essential, and it is recommended that existing proposals for parameters of quality control are verified. Indeed, even at present physicians are required by law to contribute to such quality control.

It has recently become possible for highly qualified test providers to carry out early ultrasonography which allows for the detection of more than 80% of severe malformations, heart defects and chromosomal aberrations (in number or structure) at week 13 of pregnancy [8]. These advances allow for a complete diagnosis before the end of the 22<sup>nd</sup> week of pregnancy as well as sufficient time for interdisciplinary, psychosocial counselling and the decision-making process for affected couples. All these factors combined should help to contribute significantly to the prevention of late terminations.

#### **4. The curative potential of PND should be acknowledged in public discourse**

The societal discourse on PND is too focused on the issue of “selection” while neglecting its curative potential.

The early detection of developmental disorders and high-quality cures for the fetus and the pregnant woman are part of the guidelines of the German Medical Association (Bundesärztekammer) regarding PND [1]. Improved imaging technologies have widened the offer of therapeutic approaches for pregnant women and the fetus. Endeavours to treat the fetus as a ‘patient’ go back more than 30 years. In general, a distinction is made between non-invasive and invasive methods with a curative potential.

Noninvasive methods such as angiodynography enable the evaluation of organ and heart development of the fetus as well as the detection of disturbances of placental metabolism and the cardiovascular system. Placenta praevia and Vasa praevia, for example, are conditions in which the placenta or umbilical cord vessels are positioned in front of the birth canal. If undetected, such pregnancies can put the prospective child's life at risk, or lead to remaining brain damage of the child. If the pregnant woman is at risk due to conditions such as preeclampsia, she will have to give birth at the appropriate time to prevent stillbirth or the life-threatening HELPP syndrome. Also the pharmaceutical therapy of pregnant women with a foetal arrhythmia or androgenital syndrome has become a standard treatment.

Invasive methods such as ultrasound-guided intrauterine puncture, the insertion of catheters or fetoscopy can in many cases prevent serious risks to the fetus and initiate prenatal therapy. These include intrauterine blood transfusion for foetal anaemia, rhesus incompatibility or *Erythema infectiosum*.

In the case of twin pregnancies with feto-fetal transfusion syndrome fetoscopic laser photocoagulation of the placenta is the current standard method used.

In the case of conditions such as congenital diaphragmatic hernia, parturition in one of the few specialised centres is essential notwithstanding advances in intrauterine therapy. The method of intrauterine paracentesis and catheter insertion can prevent the loss of foetal kidney function. It is also possible to close a spina bifida by intrauterine endoscopy. Fetal valvuloplasty, however, has a low success rate.

In addition to psychological support, all therapeutic approaches require interdisciplinary teams for prenatal, perinatal and postnatal care. Therapeutic aims should be clearly defined.

Prospective parents should be informed about the risks involved, which are often serious, as well as any therapeutic limitations to safeguard patients' informed consent. Interdisciplinary clinical ethics committees should also be involved in the process [2].

Prenatal diagnostics facilitates the early detection of risks to the fetus or the expectant mother and contributes to an optimised interdisciplinary supervision of pregnancies. By way of adapted pregnancy care including for parturition at the appropriate time in specialised care centres for neonatology, paediatric cardiology and surgery, newborn babies and mothers are supported towards a healthy pregnancy. As a consequence, it appears unjustified to label prenatal diagnostics as a generally 'harmful practice'.

## **5. Communication between physicians and pregnant women must be improved**

Prenatal diagnostics is a multifaceted field of practice. Test results may refer to probabilistic correlations only, and the concrete effects of a fetal disease, including possible disability, are associated with a high degree of uncertainty.

Ultrasound diagnostics as a practice is characterised by the direct contact between the physician and the expectant mother. This means that a positive test result is communicated not only via imaging and the technology as such, but usually also via the physician's facial expression. It has been shown that the physician's way of communicating results has a substantial influence on the decision-making process of pregnant women. The physician's reaction thereby presets this process at least in part. In this situation, the medical professional is confronted with the difficult task of managing uncertainties while having to handle insecurity.

When potentially relevant findings are communicated to a woman, the distinction between probable results and secure diagnosis is often difficult to understand.

An enlarged nuchal fold or so-called soft markers, although in most cases harmless changes due to the ultrasound scan, may lead to a perception of being at increased risk for Down syndrome for a concerned woman.

Physicians tend to counsel for additional diagnostics also because of their own anxieties around professional liability. Relief from concern and ongoing uncertainty cannot always be separated and require significant sensitivity from the individual treating a pregnant woman in what can be a shocking and immensely stressful situation. It is not uncommon that gynaecologists refer women to physicians specialising in PND if their test reveals any findings, without however disclosing the reason for the referral immediately.

In this way, physicians may try to minimise uncertainty for pregnant women, but accept that they might later face increased pressure of decision-making. This should be seen in the context of the health care system more generally, in which communication is considered crucial, and yet remains undervalued for the most part.

How can we conceive of a more appropriate interaction? Which factors influence communication? At which point is medical expertise asked for, and at which point primarily empathy and emotional support? What are the limitations of medical professional performance and care? How can physicians be enabled to reflect their own individual communicative behaviour? How to deliver bad news? How to communicate prognoses, probabilities, and uncertainties? In recent years, courses on delivering bad news and on risk communication were introduced into university curricula and are emerging as part of advanced job training, however often focus on decision-making at the end of life. They are also urgently needed for decision-making at the beginning of life.

## **6. PND involves decision-making processes which can be conflictual**

Pregnancy care services and PND have become increasingly intertwined. Nowadays, all pregnant women face the issue of PND through either advertising, personal contacts with users, the internet, proposals and recommendations of physicians, or even directly via an examination at the beginning of pregnancy. Examinations are often considered as part of querying whether expectant mother and child are indeed well. This concern can be connected to the desire of optimum health for the child and a willingness to accept additional costs for potential examinations. The aim of any particular test and the knowledge to be gained from it, as well as potential consequences, are often not or not sufficiently known. As a consequence, questions on how to deal with the findings of any test may arise only after PND was performed. When women are unsure whether a particular PND on offer is the right choice, they predominantly decide to eventually undergo the test.

The limited time available for gynaecologists to convey a wealth of information around PND, in part due to economic reasons, leads to a problematic situation that can be reinforced by concerns about the potential of further counselling to increase insecurity on the part of the women (cf. thesis 10). This combination of factors can tempt physicians into leaving such further counselling to the time after the test is performed.

Comprehensive counselling is, however, a prerequisite for a broad scope of decision-making opportunities. A key question used as a parameter to establish the intensity of counselling is whether potential findings may cause a decisional conflict for the expectant mother.

It has been shown that medical and psychosocial counselling prior to making use of PND bears resemblances to counselling in the case of decisional conflict.

In general, these questions should be discussed also by both experts and the public since they transcend the individual's situation. While the decisional conflict in an emergency situation is centred around the question whether a woman is able to live with and care for a child as things stand, prior to making use of PND this shifts to the issue of what an expectant mother should know of her prospective child to be able to accept it. This means that independently from any pressures and conflicts involved, the relational aspect is becoming more prominent. The aspect of acceptability and the relationship between expectant mother and prospective child is even more pronounced if PND/NIPT is chosen with the intention of excluding chromosomal alterations (such as trisomy 21). A potential termination of pregnancy is therefore always considered as a part of employing these methods. Such a potential for a decisional conflict following PND should be made explicit during responsible counselling, and is of particular relevance given the societal taboo surrounding disability and death.\*

## **7. The potential impact of PND on late terminations of pregnancies should be considered**

In 2014, 584 late-term terminations of pregnancy (post 22<sup>nd</sup> week) were carried out in Germany. Statistics, but not detailed reasons for terminations, are registered by the Federal Statistical Office of Germany (Statistisches Bundesamt).

In reality, pregnant women are often confronted with a recommendation to terminate the pregnancy should PND reveal relevant findings. As an illustration, we may consider the case of Ms H. during whose pregnancy a heart defect is discovered via ultrasound scan in week 21. The physicians in charge insist on performing an amniocentesis to understand whether the heart defect is treatable. The subsequent analysis reveals that the prospective child has trisomy 13. This result causes shock, commotion, and desperation. The family reports that they were not pressured into considering a termination, but that it felt as if the physicians were presupposing the parents would want a termination in any case, and this was going to be the only acceptable decision. The family also reports,

however, that the medical professionals changed their attitude completely when they decided to continue the pregnancy, and that all of a sudden they were treated with enormous respect – almost as if they had made a ‘heroic’ decision. Ms H. was then trying to enjoy a carefree pregnancy, and the baby appeared to do well. The delivery was without any complications, but their daughter did not survive beyond 17 days. For the parents, it was an intense and precious time that they remember also through photographs and the plaster foot prints of their daughter on the wall.

Although this is the experience of a single family only, one may ask whether, more generally, affected parents receive sufficient information enabling them to make a truly autonomous and informed decision. It is common that the medical prognosis in relation to the expected disability of the child is rather uncertain and there is no general answer to the question whether disability can or should be accepted, both individually and for society at large.

Terminations of pregnancy from week 14 onwards have to be induced like an artificially induced delivery. Since it is possible for children to survive outside of the womb from week 22 of pregnancy onwards, for terminations at this point the fetus is usually terminated through injecting potassium chloride (feticide). For affected women and families, as well as medical professionals, this is an extremely difficult situation; also because in hospital wards close-by professionals are often trying to support the survival of very premature babies.

Medical professionals supporting late terminations argue that feticide prevents enormous suffering of a future child. Prospective parents fear, understandably, that their child will be in pain and may die an agonising death from asphyxia. These concerns are usually exaggerated, however, since pain and dyspnoea are treatable in newborns. Parents’ concerns may be considered in the sense of an advance directive via a palliative care plan, both before and after delivery. In fact, many parents are able to cope with this extremely difficult situation with the help of experienced counsellors and support, although they may not have thought it possible. In retrospect they tend to report intense sorrow, but also joy and gratitude in relation to the time spent with their child. They are convinced that this support helped them to come to terms with their loss.

The aspect of palliative care should be considered more intensely, since diagnostics prior to and after birth cannot circumvent all potential confrontation with disease, death and sorrow.

## **8. A strict legal ban on late terminations does not address the complexity of individual case decision-making**

The statistics for late terminations of pregnancy in Germany record all terminations after week 22 of pregnancy due to medical or social indication without being limited to a specific time-frame (In 2014, 584 late terminations were recorded). The statistics record

all cases of high-risk pregnancies for the mother's health and well-being including cases in which the risk would have been caused by the birth of the prospective child such as cases in which the child is expected to be severely disabled (cf. paragraph 218 a, section 2, German penal code (Strafgesetzbuch – StGB)).

Until 1995, this law included the so-called 'embryopathic indication' that refers to the lawfulness of terminating a pregnancy only until week 22. Even though this former rule was confirmed as reflecting the German constitution, it was removed from the law and subsequently became encompassed in a wider margin of medical indications for terminating a pregnancy. The law remains, however, controversial.

The abolition of the explicit embryopathic indication obscures the reasons for legitimising terminations of pregnancy. Simultaneously, the discontinuation of a limited period of time for lawful terminations implies being faced with ethical dilemmas such as the de facto possibility to terminate a pregnancy if the embryo is severely diseased up until very shortly before birth. Does the presumed right to life of the embryo increase to such extent at week 22 of development that a termination from this point in time onwards should be illegal? Is it possible to justify a definite, non-negotiable right to life of the embryo by reference to the criterion of its ability to survive outside the womb and thus to justify a legal ban on termination, including against any substantial interests of the woman? While this reasoning appears inconsistent, also a potential ban on late terminations appears ill-conceived since there are cases in which the continuation of a pregnancy beyond a certain point would create unacceptable risks for the woman.

Two broadly defined groups represent the focus for this debate: the unborn with a scarce chance of survival and the viable unborn likely to be severely diseased. While the first group of cases is often discussed with reference to preventing 'unnecessary' suffering which may be employed to justify a feticide, in the second group of cases the attention is centred around risks to the women's health.

The fact that an unacceptable burden may be placed on the woman should not be overlooked. The reasoning behind the acceptability or unacceptability of preventing risks to the health of the women through a termination of pregnancy as hinted at in the legal wording of article 218 a, paragraph 2 (StGB) should be made clearer than it currently is.

A rigid time-limit would increase the pressure to make an early decision, and may in fact be counterproductive in terms of safeguarding embryonic life. The process of decision-making in this context requires sufficient time, which is already sparse.

The uncertainties of clinical practice as well as the disingenuousness of the provisions of article 218 a (StGB) suggest the need for reform, although this should not be realised via an absolute ban on late terminations.

## **9. Ethics committees cannot solve the dilemma of late terminations**

Late terminations of pregnancy are ethical dilemmas and as such for everybody involved extremely burdensome situations. This might suggest that a dedicated ethics committee would be best placed to make the final decision.

Austria, Denmark and Germany did indeed establish different approaches for the management of these situations. From the German perspective, proposals for reform have been hampered by the lack of knowledge concerning the precise reasons for late terminations as registered by the Federal Statistical Office of Germany (*Statistisches Bundesamt*). While in Germany the indication for a justified termination of pregnancy is the responsibility of individual doctors, in Denmark and Austria the decision is arrived at by involving a larger group. Both countries recognise an explicit 'embryopathic indication', which is limited to 22 weeks of pregnancy in Denmark and without time-limit in Austria.

In Austria, decision-making in relation to late terminations after week 22 of pregnancy proceeds on the basis of a so-called "consensus statement". This statement specifies that in some instances, late terminations are unavoidable and that feticide is considered the preferable form of termination to be offered. A team with interdisciplinary expertise is tasked to decide about the indication for termination. This decision then becomes binding inasmuch as other hospitals tend to accept the decision of the hospital and would not initiate a separate examination in the case of a renewed request by the woman. This is a questionable practice inasmuch as there is no legal basis for it.

In Denmark, on the other hand, the law prescribes that decisions after the end of week 22 of pregnancy are to be made by a committee, membership of which in terms of professional background is also prescribed by the law. Women are only allowed to seek a termination following a positive vote by the committee. This possibility is restricted by a general time-limit up until week 22 of pregnancy.

Is either of these approaches transferable to the German context? In other areas such as PGD an ethics committee is involved in decision-making. Even in PGD, the shifting of competencies and responsibilities involved may appear problematic. Differently from decisional conflict regarding pregnancy, however, in the area of PGD there is no comparable time pressure for decision-making.

A neutral commission's authority over the final decision may seem particularly burdensome or disempowering to the woman. She would have to accept the commission's judgment on aspects of acceptability or unacceptability for her individual case and, following the current legal and regulatory conception, on risks to her health. In this very challenging personal situation of advanced pregnancy she would appear to be primarily in need of a trusting relationship to physicians and counsellors. A potential 'outsourcing' of final decision-making authority to an external committee may, however,

relieve the pressure for doctors. This can also be achieved via interdisciplinary case counselling which could simultaneously offer comprehensive and individually tailored counselling and support.

An ethics committee, in contrast, rationalises the highly individual and quasi-impossible decision because the committee would primarily be requested to consider such cases due to its decision-making power rather than to support ethical reflection.

## **10. PND is channelling insecurities and fears and may thus appear to offer a comprehensive coping strategy**

Pregnancy, childbirth and the postpartal period are characterised by intense physical and social change and related insecurities. These centre around the potential threat to the women's and child's health and life. The changes taking place indeed affect all areas of life, since preparation for a (further) child involves considerations with regard to the partner and the social network in any given case, as well as around professional development and securing a livelihood.

As part of the medicalisation of pregnancy, childbirth and postpartal period these insecurities are translated into medical risk which, on the one hand, seems to help make these more calculable and on the other hand appear to enable the reduction or avoidance of risk via medical interventions. In turn, these offers are requested by prospective parents, which causes a forensic risk on the part of obstetricians they themselves confront through a proactive, interventionist attitude. The translation of insecurity into medical risk might, however, involve a decoupling from wider social and solidarity based aspects in this period of life.

Certainty and reassurance can hardly be achieved through the use of PND (*cf. thesis 2*). Even if a particular diagnosis may appear secure, its individual and social consequences must remain unclear. Apparent certainty as created by learning about a particular finding (for example, a genetic finding) can also lead to uncertainty in relation to additional findings. As a consequence, questions concerning prenatal diagnostics are treated as questions regarding the affected individual's values [1] in the light of specific interests. This does, however, require support. With regard to the psychological and physical limitations of the individuals concerned, there should be opportunities for becoming conscious of both diffuse and more tangible anxieties and uncertainties and relate these to one another. The more the pregnant woman can rely on a stable background of social security, the easier confronting the insecurities inherent in pregnancy becomes, and the lower the risk that such insecurity becomes projected onto prenatal diagnostics and a simultaneous hope for relief to be obtained from it.

It remains the case that the societal and socio-political regime is tasked to avoid the internalisation of PND as a comprehensive instrument to provide relief from insecurities and fears related to pregnancy and birth. PND is unsuitable both as a coping strategy

towards the control of transformative processes or as a compensation for instable social situations, and therefore requires a more appropriate contextualisation.

## **11. Psycho-social counselling involves a wide range of offers and should be promoted earlier and more pro-actively**

Throughout Germany, there is a comprehensive network of pregnancy counselling provided by different funding bodies, which work with a concept of integrated psycho-social counselling and concrete provision of support. Their services are free of charge and can be used anonymously. All women and men have a legal right to receive pregnancy counselling. In the perception of the public, however, this task of counselling is frequently reduced to counselling in the context of a potential termination, notwithstanding information campaigns online, in print media as well as a variety of events to help change this situation. The intended services do, by contrast, comprehend the offer of counselling with regard to all issues related to family planning, pregnancy and parturition, support during the first years of the child's life, as well as following the loss of a child in stillbirth or miscarriage or after the termination of a pregnancy. Pregnancy counselling services are thus available throughout the pregnancy and until a stabilised life situation has been reached.

Prenatal care as well as PND also form part of this range of counselling and support services. Pregnant women are usually aware of psychosocial issues in general and in particular in the context of PND even before their first conversation with a physician – simply as part of the confirmation of pregnancy – and these are also documented by the medical professionals involved. The opportunity to use psychosocial support services is, however, rarely explicitly referred to or indeed recommended by physicians. This may be caused by the desire to prevent any suggestion of a psychological or social deficit. Yet, it also means that physicians may miss opportunities to install psychosocial counselling in a preventative manner as a form of possible support and to make use of it also prior to PND.

All concepts of psychosocial counselling allow for an active, early and integrative approach which includes offering active consideration of PND for every pregnant woman. The relevance of these issues is evident in particular during the early phase of pregnancy, i.e. until week 14. Making use of the broad offer of psychosocial counselling usually provides relief of distress even after the first conversation. The services provide a neutral and safe environment in which the women feel that their concerns are appreciated and emotional conflicts can be considered. This possibility, in addition to the direct counselling provided, is regarded as very positive. The fact that psychosocial counselling services can be consulted also subsequently contributes further to relief of any potential distress.

Early access to these services facilitates support that can be tailored to individual needs and changing concerns – such as for example with regard the women's relationship;

education and career; income and housing; in the case of unclear findings; preterm birth or peripartum depression – and activated at any time. It is known that the majority of women confronted with findings during prenatal diagnostics would have preferred to make use of psychosocial counselling prior to PND.

In conclusion, there is a need for more proactive and systematic awareness campaigns for psychosocial counselling that can facilitate comprehensive reflection and support without reducing these to medical considerations. So far, legal provisions by themselves have not lead to increased referral or uptake.\*

## **12. Decisional conflict following PND is an irreducibly individual experience and can lead to an ‘unbearable decision’**

*“It was not clear to me from the beginning that I would have a termination. I thought to myself: my child is growing up; I can feel it. I wouldn’t have been able to terminate it. I didn’t want to take a decision because I didn’t know what to do. I wasn’t doing well at all. The physicians were very reluctant with regards to the chances for survival and healthy development of the child; they were unable to give us any reassurance. The child could have been deaf, or blind, or completely healthy. I figured that since the physicians were presenting us with the possibility of a termination, then there must be a justification for this. Against the background of how the situation was presented to us – that our child may be severely disabled and that the parturition may be very risky due to the large head – I concluded that a termination was the least risky solution.” [1]*

Pregnant women experience and handle PND in different ways since the processes of pregnancy differ. There is, however, an undisputed societal pressure to ensure the health of the future child. The situation can become extremely difficult if, following positive findings in PND and periods of waiting and hoping, a disability is confirmed and the woman is suddenly confronted with the question of whether to terminate the pregnancy she desired. At this point, at the latest, she is in an extremely difficult situation and even a decisional conflict, which is characterised by shock and trauma and hardly allows for reflection and coming to terms with the situation. Time and manner of diagnosis, as well as its context, have a substantial influence on how the subsequent period is approached. In general, there are the four options of either continuing or terminating the pregnancy; in the case of very unfavourable prognosis the continuation of the pregnancy until the near-certain death of the child; and in a few rare cases the possibility of fetal surgery. Parting with the desired healthy child and coming to terms with the loss are unavoidable in all these cases.

The decisions made affect the future, while the concrete effects are difficult to assess and it can seem almost impossible to make the ‘right’ decision. The decisional conflict is characterised by questions concerning the quality of life of the future child and the ‘justifiability’ of its life, dependent on the prospective mother’s quality of life as well as the confrontation with a ‘further loss’ due to the termination. While feeling co-

responsible, the women's partner also feels limitations and often does not see himself as in the focus of decision-making due to the absence of a physical relation to the prospective child. For the woman, in turn, there is no possibility of passing on the responsibility for the decision to be made – she is the final adjudicator [2]. She is forced to take a decision and must bear all and any consequences. Women are often concerned that their relationship could fail, and that there will be a lack of support, and are thus anxious about a future life in social isolation, faced with excessive demands. They have to individually evaluate the perspectives offered through either continuing or terminating the pregnancy as well as the question how the decision can be integrated into their plans for the future.

### **13. Bearing the responsibility of decision-making in relation to PND subjects pregnant women to substantial emotional strain**

When using internet platforms on the subject of PND [1], the focus for pregnant women is not on receiving information and evidence, but rather on the emotions involved in coming to terms with the decision-making process: *“Could somebody encourage me to handle the situation in a more relaxed way?”* The decision-making processes involved rather than PND as a medical-diagnostic practice in itself are felt to be problematic. In these communities, decision-making is often regarded as highly individual: *“Naturally, everybody needs to decide this for themselves!”* Consequently, decisions are approached in an intuitive and emotional manner, represented by expressions such as *“[...] listen to your heart”*. Desperate users expect the online community to provide emotional support in the process of decision-making: *“Please please please help me – I want to make the right decision.”*

An issue which often raises concerns is the certainty of test results in PND, which illustrates the difficulty of both measuring and weighing risks: *“I wonder, nonetheless, how certain the PND examinations and tests are?! Can you really rely on the tests?”* Feeling reassured can become synonymous with feeling at ease, both of which are equated with PND: *“[...] I can recommend doing it although it hurts but afterwards you will feel a little more reassured that your child is healthy; even if physicians cannot exclude everything you will feel better.”*

A simplified or even wrong reception of medical and diagnostic evidence, forming the base of conclusions on risky situations, can, however, be problematic. Depending on individual experience and the intelligibility of relevant information, medical results and calculations of risks may be interpreted very differently: *“You do have 3 healthy children; I cannot imagine that only because the third one was born one year ahead of ‘being under 35’ and now you are 36 there should be any radical changes.”* At the same time, the forum user's focus on certainty and reassurance reflects the influential public discourse on certainty and reassurance in relation to PND (*“I would really like to have some reassurance”*).

Recommendations that may appear paradox at first glance such as to make decisions in an intuitive manner eventually might be regarded as consistent reactions to the inextricable uncertainty evoked by PND (*cf. thesis 17*). The very same uncertainty is formally treated as a moral or ethical issue neglecting, however, a more detailed consideration of the shape and content of the problem and its implications.

These results suggest that the necessity of making a decision in the context of PND in which processes of calculating risk are highly complex and difficult to understand can cause substantial emotional distress in pregnant women. Women are often left alone in such emotional emergency situations brought about by the desire and at the same time impossibility to make the 'right' decision (*cf. thesis 12*).

#### **14. Provide ongoing professional support for women and couples following PND, independently from the decision taken**

The availability of professional counselling both in an acute decisional conflict following PND and subsequently is indispensable for women and couples to support individual decision-making. A study by the Federal Centre for Health Education (*Bundeszentrale für gesundheitliche Aufklärung*) reports that women making use of available counselling following positive findings during PND are largely satisfied [1] but also suggests increasing the availability of services. Gaps in available knowledge, the tendency to treat termination of pregnancy as a taboo as well as any other prejudice and concerns that may contribute to limited uptake of counselling for both women and couples should be reduced. Standards for the quality control of psychosocial, genetic and medical counselling in the context of prenatal examinations prepared by *Netzwerk Lebensbeginn* provide important criteria for comprehensive professional counselling in case of a pathological result from prenatal diagnostics, independently from whether the woman decides to continue or terminate the pregnancy [2]. If the woman or couple decides to terminate the pregnancy after having established a relationship to the child this decision often proves traumatising. The aspect considered most important in getting to terms with a termination is the process of grief. This process is sometimes accompanied by particular rituals of parting and mourning. It is also known that the availability of information about possible funeral arrangements and psychological support are very helpful. In addition, it is important to create a positive outlook for the future. Treating a termination as a taboo instead often has a very negative impact on the process [3]. The possibility of speaking openly about the late termination and any ambivalences, feelings of guilt, grief, or other issues as part of psychosocial support on offer is therefore crucial. The support service may be consulted directly after the termination or at any later stage. If women and couples decide to continue the pregnancy, they should receive emotional support and encouragement. The counselling offered concerning the changes in expectation about the child and future life situation should be tailored to the parents' needs. In some cases, it may be necessary to further explain the findings and/or correct wrong expectations. The future life with a disabled child can and should be planned. The provision of information

about available support, services and entitlements contribute to organisational support, and some relevant contacts can be provided already during the pregnancy. The provision of long-term and continued professional support is essential, including for the organisation of everyday life with the child.

## **15. Concern about social exclusion directly impacts decision-making in relation to PGD**

It is known that ca. 95% of all disabilities are non-congenital. Even the chromosomal aberrations trisomy 21, 13 and 18, which can be reliably detected via PND, only represent a minor part of the pool of genetic aberrations. Notwithstanding these numbers, this part of medicine is experiencing a boom period, with plenty of time, energy and resources being mobilised to advance the development of diagnostic possibilities. An increasing number of prospective parents is making use of all opportunities available to have a healthy child. According to the experience of counselling and support services, this anxiety of having a diseased or disabled child, in addition to individual factors, is due to a large extent to the limited integration of disabled individuals in society. It is well-known by most and suspected by virtually all people that real support and acceptance of disabled people have not been achieved. Although investments of 15 billion euro for the integration of disabled persons were provided in 2014, many affected parents experience their everyday life as a continuous struggle for financial and social support.

This anxiety concerning social exclusion and the tiresome struggle for everything from napkins to support for everyday care is a substantive burden for the affected families. As confirmed by a study on the situation of families with chronically ill or disabled children in Germany (*Kindernetzwerkstudie 2014*), these concerns are justified, and parents themselves are often confronted with an extremely high burden on their life situation and health. Siblings often feel neglected. In particular women are often only able to pursue a profession to a limited extent, if at all.

1 in 4 families feel that the available financial support is insufficient. For families with a low income, in particular, refunds do not cover expenses. A key criticism concerns the lack of a centralised institution that handles the entirety of questions in relation to legal entitlements (*Leistungsrecht*). In Germany, affected parents have to navigate a complex landscape of diverse institutions responsible for separate requests for support, and do not receive support for this task. This is true for all relevant requests including the identification pass for severely disabled individuals, for financial support for care, the request for additional support, the request for early educational support, for support concerning integration in a nursery or preschool, for outpatient care, leave of absence at work or part-time work, and many other issues. Responsibility for these issues lies with either health insurance or nursing-care insurance fund providers, the municipality or regional administration. Even the available offers (e.g. rehabilitation service of the German statutory pension insurance scheme [since 2002]) to support affected parents

with the administration of requests are currently still insufficiently well-known and are therefore rarely used.

Studies from Canada and Sweden, for example, show that the situation can be markedly better. Parents in these countries report being much more satisfied with their situation in comparison to German parents and indeed are receiving much more comprehensive and networked support. In Sweden, for example, every family in need is entitled to support by a professional assistant who will be available around the clock (with alternating shifts) if necessary and whose service is financed by the state.

The immense effort needed for handling everyday life in a family with a disabled or chronically ill child is a major focus for many pregnancy care services. Prospective parents are deeply concerned about a future life in social exclusion, which adds to the concerns about the diseased child and discussions about the certainty and impact of the diagnosis. Shouldering these concerns is overdue for all affected parties.

## **16. The legitimate aim of reducing the number of abortions should be addressed in its social-political context rather than through criminalisation**

Both ethically and legally, terminating a pregnancy is not a neutral act since the 'right to life' of the unborn human being has to be taken into consideration. The aim of reducing high rates of terminations is a legitimate or even politically required aim. Such a policy, however, cannot be approached via a criminalisation of the affected parties (pregnant women and medical professionals), since the decision for a termination always represents an exceptional, individual case (*cf. thesis 12*).

This is because the reasons for terminations are multifarious, and the individual burden often immense. Overall, the individual right of self-determination must remain the central consideration.

These facts need to be confronted by those who aim to reduce the number of terminations in the name of unborn human life, although it appears that in particular pro-life advocates frequently ignore them.

As a consequence, the aim of reducing the numbers of terminations can only be realised in consideration of and in cooperation with women and couples. The task at hand is of a socio-political kind which should focus on widening the margin of opportunities such that a termination does not appear to be the most obvious or even the only available option. It should address not only the issue of combining both family and professional life but more broadly providing reassurance for both couples and women as individuals that an autonomous lifestyle will also be possible in difficult future conditions. The socio-political aim should be one of creating conditions in which individual aspirations and plans do not have to be abandoned because of a (further) child but conditions in which even an

unexpected child can be integrated into these plans, and an initially undesired pregnancy may become the desirable choice.

This applies even more when the woman is expecting a disabled child. As pointed out before (*cf. thesis 15*), currently available offers of support are not comprehensive at all; financial support is insufficient; and in addition reviewing the landscape of possible help as well as applying for these services is difficult and laborious. In addition to the fact that we live in a far from genuinely inclusive society, in which disabled individuals have equal chances to everybody else, this overall outlook may create anxiety.

Only once these reasonable concerns can be confronted the number of terminations will decrease. For whoever considers high numbers of terminations as problematic, the issue needs to be tackled socio-politically.

## **17. Recent developments in PND contribute to a conflictual responsabilisation of pregnant women**

At least since the introduction of ultrasound scans in the mid-1970s, a 'subjectification' of the unborn child became prevalent, disconnecting the unity of mother and child and making the child visible and assessable. The ever more wide-reaching informational campaign around PND [*"Aufklärungsprojekt Pränataldiagnostik"*, 1] conveys the impression that pregnancy, childbirth and congenital disability can be comprehensively controlled. These developments as well as the interaction with and between other involved actors such as physicians, health insurance providers and societal expectations are changing the attribution of responsibility towards the pregnant woman. On the one hand, she is not only being pressured to behave responsibly towards herself, but also towards the unborn child. On the other hand, the mere availability of genetic tests forces pregnant women to take an active decision on whether to undergo these tests. This first decision as well as all consequential ones are becoming increasingly characterised by a changing discourse on risks, according to which, on the basis of statistical information, as of late almost any woman's pregnancy is categorised as a potential high-risk pregnancy. The individualisation of risk accompanying this development is closely connected to a societal expectation of individual risk management. Against this background, both individuals deciding against the use of any such test as well as the ones that decide to continue a pregnancy despite being aware of the risks or respectively being aware of a concrete diagnosis, are considered as being fully responsible for the consequences of their decisions. On the basis of this 'responsibilisation' it is being concluded that 'society itself' no longer has any particular responsibilities, and unconditional societal solidarity in the case of what is classified as a situation that could have been avoided – such as having a child with trisomy 21 – is therefore expected to decrease in the future.

In the case of de facto positive test results of prenatal tests and diagnostics, the majority of affected individuals experience a traumatic and conflictual situation and find

themselves in a psychological and emotional condition that makes taking a definite decision virtually impossible. Pregnant women with access to such genetic information can thus be faced with a dilemma [2] characterised on the one hand by conflicts between their individual self-determination and the assessment of what kind of life is worth living and acceptable. On the other hand, they are confronted with the more or less hidden expectations of a societal environment that negates any co-responsibility in case of a 'wrong' decision, which is increasingly often expressed in shock rather than empathy by the question: "*Were you not aware of this?*"

## **18. Leaving the introduction of non-invasive prenatal tests to commercial companies has far-reaching consequences**

Early non-invasive tests have already changed prenatal diagnostics profoundly, although these tests should be considered merely the beginning of what will be a far-reaching development. While the current focus is still on the detection of genetically caused disabilities of the unborn child, the assumption for future scenarios is that on the next level characteristics 'without any direct relation to health' [1] will be identified. From a technical standpoint, much more than this is feasible already since it was shown that the complete genome sequence of the unborn child can be mapped from maternal blood [2]. This technological development calls for political oversight and regulation. It needs to be clarified what kind of tests can be offered under which conditions and thus made accessible as well as the extent of reproductive liberty and parental 'dominion'.

If introduction and diffusion are left to commercial companies and laboratories this has far-reaching consequences. Even at present, an uncontrolled proliferation of offers can be observed, playing on regulatory loopholes and coupled to smart international marketing strategies which circumvent supposedly secure legal requirements such as the medical privilege as set out in the German law on genetic diagnostics. Moreover, the global accessibility of tests and direct-to-consumer marketing channels mean that both the erosion of the medical indication and the ban on sex selection can no longer be excluded. If political authorities fail to intervene in the interest of governability, this will contribute to a further acceleration of the current dynamics. Legal amendments to foreclose regulatory loopholes should be provided in a timely manner (*cf. thesis 21*).

Non-invasive prenatal tests represent a profitable segment of the market. Studies estimate its annual revenue at 1.3 billion US \$ [3]. The prognosis of growth amounts to 2 billion by 2020 [4]. Correspondingly, this market is characterised by competitive constraints and intense competition, including law suits around patents and other intellectual property [5]. The logic of growth of providers aims at penetrating and dominating the market globally. Apart from the US, European countries observe the highest growth rates [6]. The top positions are occupied by countries that are also currently offering prenatal diagnostics very pro-actively. NIPTs appear to allow for a seamless transition due to the already existing infrastructure and logic of the system and

are advertised merely as an additional, although profoundly improved segment of available tests. The extensive ethical, societal and individual consequences to be expected remain largely under the radar. It is the task and duty of political actors to address these consequences, and to draw societal attention to them. These actors are challenged to take an active role to curb the continuing privatisation of prenatal diagnostics.

The most recent trend on the horizon is the comprehensive introduction of non-invasive tests as so-called 'first tier tests' and thus as a replacement for the hitherto common first trimester combined tests. While high costs initially provided an important barrier against the routine use of NIPTs a solution to this issue appears to have come into view as companies such as Lifecodexx have started advertising the launch of their new qPCR-based prenatal tests claiming "cost-efficiency" and "extremely short transit times" [7].

The basis for a mass implementation of non-invasive prenatal tests has thereby been prepared. Politicians are urgently called on to counteract this development and question the rationale, significance and implications of such routine and comprehensive screening.

## **19. Neutral information about the most recent non-invasive prenatal tests is insufficiently accessible**

The internet functions as a source of medical information for many potential users, increasingly including also pregnant women. Internet-based search for information about NIPTs offers three principal possibilities: commercial businesses advertising their services in the guise of providing information (*s. thesis 20*); internet fora for the exchange of views with other affected individuals (*see thesis 13*); or neutral information offered by public institutions.

The latter should be offered by actors free from pursuing commercial or ideological interests. In Germany, the Federal Centre for Health Education (*Bundeszentrale für gesundheitliche Aufklärung*) is tasked with this informational service. Such information on the internet is, nevertheless, inaccessible.

The agency provides a flyer on its website [1], offering information about prenatal diagnostics [2]. The flyer does, however, only deal with the methods used in non-invasive prenatal diagnostics. NIPTs are not included, since the leaflet was published at a time when this new generation of tests had not yet reached prenatal care. The same is true for the 48-page strong and thus considerably more detailed booklet the authority provides on the same topic [3].

These publications were prepared in cooperation with a number of charitable organisations: Workers' Welfare Association (*Arbeiterwohlfahrt*), Caritas (Catholic welfare association), association of social movements *Paritätischer Gesamtverband*, German Red Cross, Diakonie (social welfare organisation of Germany's Protestant churches), and *Zentralwohlfahrtsstelle der Juden in Deutschland* (social welfare

organisation of the German Jewish community). As a result, all contact points offering counselling for pregnant women refer them to the very same source of information. The Federal Centre for Health Education also offers information about the new generation of blood tests for chromosome malformations on the internet platform *familienplanung.de* yet it is difficult to track down [4]. The main platform of the Centre provides very detailed information on the topic of NIPTs [5]. However, this offer is addressed to an expert audience, which shows in the terminology used such as “gender test” and “family balancing”.

Looking for other offers on the internet does not prove very successful either. The website of an association offering pregnancy counselling even refers to the term “prenatal blood test” on its landing page without however providing further information. Yet, the same association did offer a newsletter on family planning and developments in prenatal diagnostics with a focus on NIPTs in May 2014. While providing comprehensive information about NIPTs, it is also addressed to an expert audience.

A more intensive inquiry yields numerous results from the commercial sector and some information for an interested expert audience. Affected women, of all people, miss out, and presumably end up in internet fora in which medical facts are not being taken into account.

Health institutions, in particular the Federal Centre for Health Education, are thus urgently called on to make their informational resources on the topic of NIPTs available not only to experts but to all interested parties – especially all pregnant women – and if possible contemporaneous to medical developments. Nowadays, affected individuals often consult the internet before proceeding to face-to-face counselling. Hence, it is necessary to provide neutral information on the internet to allow prospective parents to make an informed decision.

## **20. An information policy motivated by commercial interests undermines rather than supports informed decision-making**

The dominance of commercial companies and providers in the sector of non-invasive prenatal tests does not only affect the dynamics of the market but also the accessibility of neutral information. The strong presence of globally operating enterprises on the internet more or less inevitably results in women seeking out information on the websites of these commercial providers. The booklets and flyers available from these providers do, however, contravene the principles, standards and quality criteria of informed decision-making.

When financial interests are the priority, it is evident that no company would present its own offer in a neutral, balanced or critical way but quite simply as the best product available. Nonetheless, the basis for any informed decision can only be a balanced assessment of all relevant aspects of any diagnostic method including advantages,

disadvantages, possibilities and limitations. To enable them to make an informed decision, women also need to be informed about their rights, including the 'right not to know', as well as available alternatives.

These quality criteria are not met by current information policies of commercial providers. This was already demonstrated for the anglophone area [1], and also became the subject of an internet search and analysis for the German speaking area in the context of the discourse project [2]. The results demonstrate the following deficits:

1. *Lack of evidence*: Figures referring to reliability and significance or explanatory power of tests are not substantiated but only asserted. How and under which conditions of study the figures were established remains intransparent.
2. *Lack of information on the 'right not to know'*: All the leaflets examined suggest that knowledge and reassurance are desirable goods which are definitely achievable via the tests offered. The right not to know is being passed over, and the potential for decisional conflict or dilemmas connected to the knowledge gained is not being mentioned. Anxiety and psychological stress that may accompany the period of waiting – due to the possibilities of choice as well as the ensuing responsibility – are not being broached.
3. *Concealment of consequences of selective approach*: The potentially selective impetus of prenatal testing is being concealed. Terms such as 'termination of pregnancy' or 'abortion' do not appear. In contrast to the message that these tests can be carried out early on in pregnancy the websites consistently depict women at an advanced stage of pregnancy with their hands lying protectively on their baby bellies. These are coupled with messages such as "Now that you are pregnant, nothing is more important to you than supporting the healthy development of your child" [3], creating an image of protection and care as well as appealing to the women in their role of responsible expectant mothers.
4. *De-contextualisation and ethical neutralisation*: The ethical framework prenatal diagnostics relates to is completely disregarded. Isolated from their context, the tests appear as ethically neutral and 'pure'. Putative characteristics such as harmlessness, absence of risk and the simplicity of taking a blood sample are foregrounded. The central argumentative strategy is framed in terms of the contrast to and comparison with the miscalculations and disadvantages of previous methods. The argument presented is part of the logic of the market and serves to stabilise the logic immanent to the system.

The overall upshot is that a balanced and quality-orientated information policy of commercially independent actors external to the system is urgently needed.

## **21. Effective regulation of non-invasive prenatal tests cannot be achieved within national law only**

Without any doubt, non-invasive prenatal tests (NIPTs) offer an advantage compared to conventional methods of prenatal diagnostics. Via a simple blood test, a screening of the unborn child has become possible, without submitting the pregnant woman to risks of infection or even the dangers of a miscarriage due to the intervention such as during amniocentesis. While blood tests therefore offer low-threshold prenatal diagnostics they also represent a new regulatory challenge. Since blood samples can also be taken by non-professionals and are easy to ship, it is possible for commercial providers of NIPTs to sell such medical products as direct-to-consumer tests (DTC tests) to couples who desire a child.

While offering direct-to-consumer tests contravenes the German law on genetic diagnostics, the regulatory situation in other European countries is heterogeneous [1]. A market analysis shows, moreover, that NIPTs can already be ordered directly via the websites of foreign providers. The providers operating in this market do not only use marketing strategies that trivialise the procedure but in part are aggressively targeting women without any health issues (*see theses 18; 20*). The currently available offer of genetic tests as well as the marketing strategies observed demonstrate that some of the companies aim at offering direct-to-consumer tests. If this approach were to ensue, this would mean that prenatal testing and the responsibility for it would shift from the medical context towards pregnant women, circumventing the obligatory genetic counselling as stipulated by the German law of genetic testing as well as the privilege of the medical profession. Moreover, compliance with quality standards, such as in relation to the handling of genetic information by foreign laboratories, cannot be guaranteed.

The future development of NIPTs is also likely to be accompanied by foreign companies offering tests that promise to screen for factors banned by the German law on genetic testing (such as for late-onset hereditary disease). It is difficult to foresee at this point whether such blood tests would motivate women to 'test pregnancies' but a number of ethical challenges can be inferred from the experience of DTC tests [2]. The information made available by commercial providers is insufficient in many cases [3]. The quality of genetic counselling, if taking place at all, cannot be guaranteed and the experience with current DTC tests does not contribute to sufficient trust in the integrity of genetic data processing and storage. In addition, the insufficient provision of information to users has created new challenges for conventional genetic counselling as well as for doctors, whose expertise is still called for following uncertain results of NIPT [4].

Current national law-making and guidelines are inappropriate for regulatory intervention concerning the market of DTC genetic tests [5]. Against the background of existing doubts about an ethically responsible free market of prenatal genetic tests new strategies are required to close the currently existing regulatory gaps. Alongside voluntary commitments of commercial providers which should comprise guidelines for

advertising, marketing and standards for genetic counselling, also comprehensive European regulation or licensing of DTC genetic tests would seem advisable to reconcile the increasing offer of NIPTs with ethical standards.

## **22. The current discourse on NIPTs is fostering deficit-oriented views and stigmatisation of disabled individuals and thus contravenes UN CRPD**

The ratification of the UN Convention on the Rights of Persons with Disabilities (CRPD) includes a commitment of the signatories to take effective and appropriate measures to raise awareness about individuals with disabilities throughout society and to foster respect for their rights and dignity [1]. Prejudice, stereotyping and stigmatisation are to be countered and opposed.

The dominance of the market, simultaneously to this commitment, helps diffuse a discourse which feeds reductionist views of persons with disabilities. The leaflets on prenatal tests by providers [2] convey images of normality, health and disease which present disability primarily as dysfunction, abnormality and suffering. Wording and style of these leaflets conflict with the raising of awareness as required by article 8 of the UN Convention on the Rights of Persons with Disabilities, exemplified by the following aspects:

1. *Protonormalistic strategies*: The arguments used seek orientation at a norm that is strategically being re-inscribed. The point of reference is the normal status of chromosomes or “normalised value of chromosomes” [3] of human beings. Variations of the set of chromosomes are referred to with terms such as *chromosomal dysfunction* [4], *chromosomal abnormalities*, *chromosomal anomalies*, *erroneous distribution* etc. Pregnant women are called on to promote “the healthy development” [5] of their child. The counterpart of the healthy development and the “welfare” [6] of the unborn child is represented by emblematic terms such as *malformation*, *mental disability*, *weak or moderate mental and physical developmental delay*, *multiple birth defects* etc. This creates a binary opposition and difference based on the strict division between the normal and the pathological, of healthy vs diseased, and of disabled as anormal and diseased.
2. *Discourse on risk*: terminology and means of representation focus on risks and dangers. Risks are overemphasised and contextualised with reference to a double meaning: 1) as the risk of an undetected chromosomal dysfunction, and 2) as the risk of an unnecessary miscarriage following an invasive procedure. Both of these risks are being associated and summarised in the short formula that seems to provide a solution to the

dilemma: “Get reassurance, without any risk for the child” [7] Protective factors during pregnancy – such as for example the creation of a secure bonding with the unborn child – are masked. As a result, an unrealistic perception of risk is created as part of which the anxiety to have a child with a disability is increasing. The imagined child is represented chiefly as a risk.

3. *Medical reductionism*: wording and style of the materials convey a medical-reductionist view of disability. The biological basis – a change in the set of chromosomes – is equated with disease and disability. Social context factors, general societal conditions, barriers and influences are being masked. The meaning of disability created is biased, reductionist and retrograde as against the awareness and knowledge anchored in UN CRPD that disability results from the interaction between persons with impairments and attitudinal and environmental barriers. While the medical model of disability finally seemed superseded in history with the ratification of UN CRPD the discourse on NIPT as dominated by the testing industry virtually re-introduces this model through the backdoor.

## **23. Ethical debate on fundamental questions, although resource-intensive and not always leading to unambiguous results, is indispensable**

An old joke suggests that when two lawyers discuss, three opinions emerge. This joke also applies to ethics and philosophy, even though it would have to target not merely the pluralism of views but also differing terminologies, methods and identities. Within professional ethics, a wide spectrum of possible positions is being vigorously endorsed. Such a plurality or such disagreements – depending on whether this state of affairs is considered as positive or negative – entails that ethics ‘in itself’ cannot provide definite and clear-cut answers to acute ethical questions.

One conclusion to be drawn may be to look for faster and more definite answers without ethics. This thought may appear appropriate, in particular when philosophical and theological ethics becomes occupied with debating fundamental questions only, rather than taking part in concrete problem solving, orientated towards the current technological state of development. Nonetheless, precisely these fundamental debates which question issues that presumably were already ‘ticked off’, and create links to problems which initially may seem to deviate from the acute problem, are indispensable.

In relation to the rapid technological and fact-creating developments in prenatal diagnostics ethical reflection and societal debate almost inevitably lag behind as they require dedicated time and do not usually lead to definite consensus. This, however, should not be considered as a deficit to be removed but rather interrupts a logic which

would otherwise become routine business. Just as with regard to medical and bioethical issues in the past, current and future questions with regard to PND will not be resolved unambiguously in ethical terms and consistently throughout society. Nonetheless, a more or less definite and homogeneous political and legal arrangement is, and always was, necessary.

Such arrangements do, however, merely result from pragmatic necessity and do not provide definite answers to the underlying questions rather than temporarily suspending them. The possibility to pose the same questions of principle in ever-changing new contexts, caused by new developments, is not merely a necessary corrective for what might have been wrong decisions at earlier points but also forces us to account for whether what has become self-evident should indeed be self-evident.

It should not come as a surprise that urgent issues around NIPTs do not raise exclusively new questions – such as whether these developments are heading towards “designer babies” or how to regulate marketing internationally – but also continuously bring older questions that may have seemed ‘resolved’ to the fore. Indeed new prenatal diagnostic methods do not merely point to the future but also to the past – from the controversy about abortion to in vitro fertilisation. It is the purpose of debates on principles to integrate current developments into a continuum and provide a reference point for the question whether the human being in itself rather than the technology is still the central consideration.

None of this does imply that ethics should only be dealing with issues of principle; the challenge is rather to create a bi-directionality in which the consideration of issues of principle is always linked to concrete challenges and, on the other hand, all concrete attempts at solving problems can also be looked at from a distance and allow for asking fundamental questions.

## Explanatory notes

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#### **Thesis 6**

\* This thesis is based primarily on results of the pilot project “Psychosocial counselling in preparation of prenatal diagnostics at the interface with medical counselling” (2008-2011) which was supervised by Prof. Anke Rohde and colleagues ( Gynaecological psychosomatic medicine, University Hospital Bonn) and Prof. Erika Feldhaus-Plumin (Evangelische Hochschule Berlin) and commissioned by the Bavarian ministry for work, social affairs, family and integration. For an outline of the project please see: Sozialdienst katholischer Frauen Landesverband Bayern (ed.) (2015) *Beratung im Kontext der pränatalen Diagnostik. Eine Handreichung für die Praxis*, available at: <http://www.skfbayern.de/aufgaben-projekte/schwangerenberatung/praenataldiagnostik/>.

#### **Thesis 10**

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#### **Thesis 11**

\* See note to thesis 6 above.

#### **Thesis 12**

[1] The quote is taken from an interview as part of a documentary film project on prenatal diagnostics, dying and death by the Austrian philosopher and filmmaker Thomas Fürhapter.

[2] Cf. Peters JS (2011) *Spätabbruch. Schwangerschaftsabbruch nach der Pränataldiagnostik. Psychosoziale Beratung zwischen Recht auf Leben und Recht auf Selbstbestimmung* (Hamburg: Diplomica).

### **Thesis 13**

[1] The thesis is based on an explorative inquiry of the online fora *elternforen.com*; *babyclub.de*; *paradisi.de*; *rund-ums-baby.de*; *forum.gofeminin.de* and *babyforum.de* using the search terms “pregnancy” and “prenatal diagnostics”.

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[2] Netzwerk Lebensbeginn (ed). (2013) *Achtsam von Anfang an. Qualitätsstandards in Einrichtungen für Familien rund um Schwangerschaft, Geburt und frühe Kindheit*, available at: [http://www.kinderjugendgesundheit.at/uploads/Qualitaetsstandards\\_in\\_Einrichtungen\\_fuer\\_Familien\\_Bericht\\_BMfWFJ\\_Jaenner\\_2013\\_web\\_v0\\_01.pdf](http://www.kinderjugendgesundheit.at/uploads/Qualitaetsstandards_in_Einrichtungen_fuer_Familien_Bericht_BMfWFJ_Jaenner_2013_web_v0_01.pdf).

[3] Cf. Peters JS (2011) *Spätabbruch. Schwangerschaftsabbruch nach der Pränataldiagnostik. Psychosoziale Beratung zwischen Recht auf Leben und Recht auf Selbstbestimmung* (Hamburg: Diplomica).

### **Thesis 17**

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[2] Samerski S (2010) *Die Entscheidungsfälle. Wie genetische Aufklärung die Gesellschaft entmündigt* (Darmstadt: Wissenschaftliche Buchgesellschaft).

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[4] Ibid.

[5] Agarwal A et al. (2013) Commercial landscape of noninvasive prenatal testing in the United States *Prenatal Diagnosis* **33(6)**: 521-31.

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[2] Bundeszentrale für gesundheitliche Aufklärung (Federal Centre for Health Education) (2011) *Pränataldiagnostik – Beratung, Methoden und Hilfen. Eine Erstinformation*, available at:  
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[3] <http://www.familienplanung.de/service/broschueren-der-bzga/praenataldiagnostik>.

[4] <http://www.familienplanung.de/schwangerschaft/praenataldiagnostik/bluttests-auf-trisomien>.

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[2] The authors compared and analysed the following leaflets for tests: Harmony (Ariosa/ Hoffmann La Roche), IONA Test (Premaitha), MaterniT21 Plus (Sequenom), Panorama (Natera Amedes), PraenaTest (LifeCodexx), Prenatalis (Verinata Health/ Illumina).

[3] <http://www.ariosadx.com/>.

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[4] <http://lifecodexx.com/>.

[5] <http://www.ariosadx.com/>.

[6] <http://www.prenatalis.de/>.

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All links last accessed in March 2017.