THE PSYCHOLOGICAL IMPACT OF THE PRENATAL DOWN SYNDROME DIAGNOSTIC TEST

MELANIA ELENA (POP) TUDOSE[1], PETRU ARMEAN[2], VICTOR IOAN POP[3]

[1,3]"Iuliu Haţieganu" University of Medicine and Pharmacy, [2]"Carol Davila" University of Medicine and Pharmacy

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Abstract: *Background: Prenatal Diagnostics Testing is the only current method of clarifying suspicions on Down syndrome obtained as a result achieved through the increased risk screening. The pregnancy, by definition, is a period of great transformation, with multiple organic and psychological adaptation processes, while sensitivity touches high peaks and anything, as small as it could be, may disturb the equilibrium and cause psycho-emotional imbalances. Most often the diagnostic result of the screening is most likely to infirm the screening and will most probably reinstall the psycho-emotional stability, but mother's already anxious attitude may have already affected the child's future emotional development and behaviour. Methods: The present study is a narrative analysis, based on a collection of data from studies published in the databases of PubMed / Medline, SpringerLink, EBSCOhost, Elsevier and ResearchGate. Results: From reviewing the studies, there have been highlighted three themes: associated psychological effects, consequences and coping methods and practices and methods that can influence the psychological effects. Conclusion: The consequences and psychological effects, closely linked to the necessary diagnostic procedures and possible outcomes, require more interest from specialists which are now more focused on the diagnostic component.*

INTRODUCTION

Pregnancy is a natural and profound physiological stage of a woman's life which takes place in normal conditions for most women.(1) However, its completion involves a lot of investigation in order to detect in time the emergence of potential health problems of mother and fetus. One of the investigations related to the fetus is the prenatal genetic testing for fetal aneuploidy is the case for the Down Syndrome (DS) in this case, an important technological acquisition in modern obstetrics. This may bring many answers but can also generate many questions, controversial decisions and ethical dilemmas.(2,3)

DS is a consequence of the presence of an extra chromosome 21 (most common) or just supplementing critical region 21q22, both producing the over dosage gene effect. It is the most common genetic cause of mental retardation (from moderate to severe), the most common chromosomal damage of the newborn and the most compatible autosomal trisomy with survival. DS incidence is 1/700 live births.(4,5,6) Current diagnostic algorithm includes both DS prenatal stage and the diagnostic screening, specifying that screening tests relate to prediction, indicating a suspicion, while the detection and diagnosis refers to the actual certification and it is the only way to certify the suspicion.(6,7)

In order to obtain the necessary material for fetal genetic analysis, diagnostic testing uses invasive procedures such as Amniocentesis (AC) and Chorionic Villus Biopsy (CVB). CVB can be performed between the 10^{th} and 13^{th} week of amenorrhea and carries a risk of miscarriage of 2-3%, while classical AC can be performed starting with the 15^{th} – 16^{th} week of amenorrhea and carries a risk of miscarriage by 0.7%.(8,9) Conventional Karyotyping is still considered the gold standard for prenatal diagnosis of DS but has the disadvantage of a longer duration to obtain the result. Faster results can be obtained through the FISH or QF-PCR tests, but given that these are specific techniques with low sensitivity, experts recommend complete cytogenetic evaluations through the classic Karyotyping that has the advantage of 100% accuracy.(10,11,12,13). The pregnant woman's strong emotionally reactions to any potential threat of pregnancy or fetal status apply to invasive diagnostic procedures, too (AC and CVB).(14,15,16,17,18)

Associated psychological effects

Pregnant women who performed prenatal diagnostic procedures are experiencing much higher levels of stress and anxiety, sometimes with depressive disorders compared with pregnant women whose task performed within normal parameters and do not require any additional intervention for possible diagnoses.(17,18,19,20) Stress occurs since the blood is collected for screening and gets higher with achieving an increased risk and suspicion of abnormality. The decision itself to perform an invasive procedure is difficult, complex and the short time decision pressure further increases the stress levels and anxiety.(21) Anxiety before the procedure was identified by reference to invasiveness, during and immediately after the procedure with regard to child endangerment and whilst awaiting the results referring to the result.(14,15,16,17,18,20) It can fluctuate over time, without permanently disappearing and showing maximum levels immediately after the invasive procedure and during the awaiting the results of diagnoses.(21)

Most researches focused mainly on anxiety and negative emotions related to the procedure itself and possible adverse effects on the fetus or pregnancy, so there are few studies focused on the emotions experienced during the period of time for the results to be ready.(16) It seems that stress and anxiety related to the procedure itself, however, is not a clinical

[1]Corresponding author: Melania Elena (Pop) Tudose, B-dul. N. Bălcescu, Bl. Camelia, Ap. 16, Buzău, România, E-mail: melaniaelena_tudose@yahoo.ro, Phone: +40721 751 540

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problem.(17) Pending time after an AC may extend to 3 weeks, during which uncertainty related to child health intensify obsessive thoughts about possible abnormal results would entail a decisional moment of existential importance. Frequently, this period associates psycho-emotional feelings such as anger, guilt, worry, anxiety and sometimes depression, for many women, hard to cope with or to manage.(16) This situation prone to choosing extreme coping mechanisms, opposite reactions, denial or defensive reactions.(15)

Consequences and coping methods

Psycho-emotional imbalance may affect fetal-maternal attachment with possible consequences for mental and behavioural development of their child, even if subsequently refuted result.(17,20) Mother and fetus have a symbiotic relationship, so that if a disease affecting one of the two most likely will affect the other one. Psychologically, it was found that the level of stress and anxiety is much greater when the test results have altered fetal health, compared with altered test results related to maternal health. (1)

During the waiting for the results period, some pregnant women are focusing predominantly on what could be wrong with their fetus, an attitude that enhances anxiety and cortisol release. Excessive exposure of the fetus to high levels of cortisol may promote development of type II diabetes and obesity in adulthood but also learning and memory disorders, up to and including the functional impairment of the frontal lobe executive.(20,22) Generally, anxious pregnant woman is associated with negative effects both on her and on the baby, effects whose consequences may be short-termed, materialized only through an increase in symptoms related to pregnancy or intensification of vices harmful in the medium term by the appearance of obstetrical complications, including birth, as well as on long-term by stimulating post-partum maternal depression post-partum or worse, by impaired development of neuropsychiatric fetus that would become a child with psycho-behavioural issues later.(23)

Coping is a very important process in situations of stress and anxiety. There are studies that have shown that optimism and positive attitude of women attending stage diagnosis lowers stress levels and keeps the situation under control, unlike avoidant women, addressing a maladaptive attitude.(15) An example of maladaptive coping can be considered the imposition of an emotional distance up to mental pregnancy rejection of their child, mental abandonment and lack of communication with him throughout the period of waiting for the results. After receiving favourable outcome, the return to the mental status of pregnant woman attitude occurs and thus resume mother-child bond, but the psychological status of fetal damage may already have occurred.(24,20) Some pregnant women cope by mentally imposing to themselves that they bear a healthy child and that the procedure will only bring this confirmation. It is an instinctive protective attitude based on the suppression of fears that aim to avoid the feeling of anxiety. Not based on conviction, sliding to clinical anxiety is easy to be done.(17)

Practices and methods that can prevent or influence the psychological effects

For a less intense emotional involvement, women think that the diagnosis should be as early in pregnancy as possible and the information has to be complete, adequate, interactive, personalized and to take place in the prescreening period.(17,21) Both AC and associated CVB score for anxiety and depression.(16,17,20,18,19,24) Unlike AC, CVB has the advantage that it can be done earlier in pregnancy when we are still talking about embryo and not fetus and the result can be obtained in a few days.(14) But CVB is associated with a higher risk of miscarriage than AC, which causes higher levels of depression.(25) With regard to genetic analysis, it was found that providing early results significantly decreases the level of maternal anxiety. This is possible by making besides classical Karyotyping, a rapid molecular test (FISH, QF-PCR), too.(16) The Non-invasive Prenatal Testing (NIPT), considered the technological cutting edge revelation, can analyze early fetal DNA in the mother' blood, from 9-10 weeks of pregnancy. NIPT can only be used as a screening test, not a diagnostic test, confirmation of a positive result also belonging to obtaining fetal material by the known invasive procedures.(26) Using NIPT as a second contingent of screening for all pregnant women at risk (1-150) significantly decreases the percentage of AC candidates.(27)

Information and counselling even before proceedings are efficient and handy method that can help women to cope with emotional difficulties related to the procedure and concerns about the results.(15,17) Increased intake of information through the use of written materials and presentations including video to increase the level of knowledge, reduce decisional conflict and increases the satisfaction of an informed choice.(28,29) There is question whether too much information could cause an increasing anxiety if extensive knowledge about the negative implications and limitations associated tests can influence a woman's decision making and can lead to an overestimation of risk associated with the abortion procedure and an underestimation of the rate of detection.(30) The opposite pole demonstrated that anxiety enhances the minimization of information.(31).

Using Decision Support Technologies (DSTs) as supplementary educational materials that complement interactions with professionals could have an important decision not only in mitigating conflict but also in lowering anxiety. Unfortunately, it was found that the amniocentesis procedure used DSTs little compared to other medical conditions associated with decisional conflicts.(32,21,33) The use of cognitive therapy techniques adapted to the pregnancy would be useful not only to reduce the diagnostic testing related anxiety, but to an increase in psychological well-being of both mother and unborn child and the relationship between the two.(23) Emotional support and involvement of partners in the implementation of this step decreases stress and anxiety of pregnant women but not related to the procedure, too.(18)

Discussion and Conclusions

DS prenatal testing involves a variety of sensitive and specifics, as well as risks of invasive procedures, an important potential to trigger a series of consequences with long lasting effects. Definitely, prenatal testing needs to be addressed and understood as an option and not an obligation to inform women since the first recommendation of screening. Trying to spare the pregnant and not to tell that from a prenatal test screening can lead to termination of pregnancy, besides violating the rule of informed consent is a mistake that can have serious psycho-emotional repercussions over that person. The development of such a scenario must not be maximized nor ignored or minimized.(15,31,21) Failure to provide necessary information, inadequate skills processing and rendering specialists, poor communication, lack of segregation, ignorance, fears, psychological reactions adjacent informed consent, decision making and facilitating its tactics in latter are gaps and weaknesses that can cause cognitive distortions affecting the reasoning of those directly involved.(31,34)

Psychological stress could be minimized by widening maternal-fetal care team and other specialists' inclusion in providing information and psychological support, including the time spent awaiting the results of diagnoses.(20,31,35)

Prenatal diagnostic technologies are currently performing and there is a concentration of researchers carried to the extreme in this regard, but their effectiveness in practice will never be as powerful there will not be paid similar attention to issues and implications associated especially to the psycho emotionally ones.(25,18)

Monitoring pregnant women should not be purely obstetric parameters and directed only toward physical status but also an emotional assessment of the level of stress, anxiety and depression especially in covering the distance of achieving a screening and final output after invasive procedure.(15,18,25)

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