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## The Triple Test and Down Syndrome: between Feticide and Therapeutic Abortion

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### Abstract

Down syndrome (DS) or Down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. Down syndrome is the most common chromosome abnormality in humans and represents a complex multifaceted disorder with multiple medical and bioethical implications. Thereby, it is typically associated with physical growth delays, a particular set of facial characteristics and an important degree of intellectual disability. The average full-scale IQ of young adults with Down syndrome is around 50. A number of tests can be used to screen for Down syndrome, with varying levels of accuracy and invasiveness. Several blood markers can be measured that can be used as part of combined tests to predict the risk of Down syndrome. Abortion after 12 weeks and called therapeutic abortion may be performed only due to precise medical cause. Down syndrome is not included in the categories of conditions that justify interruption of the pregnancy by therapeutic grounds. Still, most parents decide the interruption of the pregnancy in the case of finding out about the probability of having a child with Down syndrome.

*Keywords:* Down syndrome, triple test, genetic disorders, education.

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## Introduction

Down syndrome is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. Historical data regarding this disorder show that the syndrome was first described in 1866 by the English doctor Langdon Down, who built the London a care center for people with disabilities. Unfortunately, L. Down used the term mongolism for this anomaly, which had a negative effect on the trisomic children and their families as well (Tobolcea 2002 ). Since then it has started a titanic work for the discovery of a treatment, realizing that new technologies will allow the antepartum detection of Down syndrome which will lead to the abortion of the affected fetuses. And in fact, in USA-today, up to 92% of fetuses with prenatally detected Down syndrome are aborted (Van Dyke *et al.*, 1995). Down syndrome is a very important health issue being the most common genetic disorder with a frequency of about 1 in 700 newborns. World wide about 7 million people have been diagnosed with this syndrome, out of which about 400,000 people live in the U.S Kent, (Kent *et al.*, 1999). In Romania there is no official statistics, but it is estimated that there are around 30.000 cases [<http://www.aspgorj.ro/promovare/anal%20sd%20romania.pdf>]

Down syndrome represents a complex multifaceted disorder with multiple medical and bioethical implications. Regarding neuropsychiatric disturbances, the chromosomal abnormality which causes the Down syndrome or Trisomy 21 is responsible for the cerebral changes and the various problems that alter the physical and mental development of these subjects. Most of these organic changes occur before birth and especially during fetal development, specifically in the last 6 months of intrauterine life (Hattori, Fujiyama, & Taylor, 2000). Trisomic subjects are situated in the classes of moderate and severe mental retardation and not in the profound mental retardation class. The level of their IQ score is frequently around 50. However, studies show that some people suffering from Down syndrome may manifest psychological disturbances and psychiatric disorders including: adjustment disorder, low self esteem, inattention, impulsiveness, autistic spectrum disorder, depression, dementia of Alzheimer type with an early onset (Capone *et al.*, 2013; Jones *et al.*, 2013; Kent *et al.*, 1999; Nicham *et al.*, 2003). Also, an overall decrease in the quality of life is seen in these patients (Geok, Abdullah, & Kee, 2013). Other clinical non-psychiatric disturbances have been identified among Down sufferers. Congenital cardiac defects, obstructive sleep apnea, hypothyroidism, digestive malformations, altered immune system function, leukemia, hearing impairment and cataract seems to have a higher prevalence in Down patients than in general population (Malt *et al.*, 2013; Patrick *et al.*, 2014; van Schrojenstein Lantman - de Valk HM *et al.*, 1996).

The coexistence of other medical disorders and a decreased capacity of work determine an increase need for medical, social and psychological support with negative financial impact for both patients and their families. Considering these

aspects and the fact that parents become more aware about complications associated with Down syndrome, finding about the diagnostic explain the great negative impact for the family. The fear of stigmatization and rejection by the community may increase furthermore the negative emotions towards having and raising a child with Down syndrome (Ahmed *et al.*, 2013). In this context, it seems that most parents decide to terminate the pregnancy. Studies have found that the number of abortion after prenatal diagnosis of Down syndrome is very high, varying from 60 to over 90 percent worldwide (Mansfield, Hopfer, & Marteau, 1999; Natoli *et al.*, 2012). However, the trisomic child does not depict a very profound disability, despite the common belief. Still, the physical and mental development disability of the Trisomic child is real and should not fade to various fantasies pretending it's normal and refusing to see it otherwise than this way (Tobolcea, 2002). Nature is the one that provides each of us, whether disabled or not, the development potential to which only some subjective limits of our body are imposed.

The medical dimensions and socio-familial impact of the issue regarding Down syndrome had greatly influenced research in prenatal medicine in search for a correct fetal prognosis and have brought in the field of obstetrics over the past few decades more tests at various gestational ages, among which the triple test, performed in weeks 14-21 of intrauterine life. This test diagnoses up to a 70% rate the cases of children with Down's syndrome, as well as two other deficiencies involving chromosomal anomalies. In this way, the delicate situation for both the obstetrician and the family is the following: in the case of positive diagnosis of Down's syndrome should therapeutic abortion be carried? - especially since abortion on request according with the legislation may be carried up to 12 weeks – or breaking pregnancy after 14 weeks means to produce a feticide due to the current possibilities in the assistance of Down's children ( Deborah *et al.*, 2009; Gronowski, 2004).

However, general knowledge regarding Down syndrome may be limited to only negative implications and public information may emphasis the problems correlated with this condition. Thereby, in recent years the life expectancy has risen, approaching the average life expectancy of the general population. For a better development to be reached, it is recommended that children diagnosed with Down syndrome to live with their families (not to be institutionalized), to participate in educational, vocational, social and recreational programs. In this way, in the United States children with Down syndrome are integrated within the public education system and there are adults with trisomy 21 who graduated from high school and have a college diploma (Pueschel *et al.*, 1997). In recent years, there are a growing number of associations and foundation supporting people with Down syndrome, but this is not the case in the developing countries.

Also, many studies have been performed in the attempt to find therapeutic ways to cure Down. In 1996 the Jerome Lejeun Foundation was created, whose

primary purpose is represented by finding a treatment for patients with genetic intellectual disabilities (in particular Down's syndrome). The Foundation has recently patented a family of molecules that inhibit one of the enzymes responsible for mental retardation associated with trisomy 21. This is considered to be cistation â synthase (CBS) which till now has been found to have 6 classes of inhibitors (Down's Syndrome Research Foundation (2005). Over the years, these Foundation researchers have proposed other possible therapeutic measures, among which the best known is the one that uses the GABA receptors inhibitors (studies have been performed on animal models) ( Down's Syndrome Research Foundation, 2005; Dykens, 2007). In this way, the FDA has already approved the use of Posiphen (an inhibitor of the synthesis of amyloid precursor protein APP) in the treatment of Down syndrome.

Currently, the most therapeutic used method in case of a Down pregnancy is the abortion.

In this context, the subject of screening for Down condition is a matter of extensive debate, since the discovery of antenatal detection tool for Down syndrome, the number of abortion caused by this genetic disorder has increased, as expected. Withal, the early detection tool may give parents some possibility of choice regarding keeping or not the pregnancy. Monitoring investigations for Down syndrome as well as for other conditions require the consent of the applicants. Some of the women believe that knowledge of the chances of having a child with Down syndrome would help them be prepared for this possibility, while others prefer not to have any information of this kind throughout pregnancy. However, it is crucial for all the pregnant women to have access to correct, nondirective, and sufficient information about prenatal diagnostics (Bjorklund *et al.*, 2012).

The process of screening involves the serum determination of various fetal and placenta compounds. The values obtained for these markers together with certain maternal clinical data (age, gestational age, weight, race, diabetic status, smoking, fetus number, in vitro fertilization) are used in a complex mathematical model (carried out with special computer programs, e.g. Prisca) in order to provide a calculated risk. The laboratory establishes a cut-off (limit) for each condition. In this way, getting a high risk indicates that the obtained value exceeds the set cut off, but does not have diagnostic value, as it rather indicates the advisability of further additional investigations. It is important to highlight that the screening tests (monitoring) do not diagnose the Down syndrome, but rather provide information about the probability of the fetus to manifest the condition. Also, performing triple test in obstetrics aims precisely to inform family and therefore has an informative role solely and does not encourage the abortion that is not included in the legal frames.

### ***The testing period***

The triple test can be carried out between the 14 and 22 weeks of pregnancy; however, the optimal period is between 16 to 18 weeks. This aspect has very important bioethical implication for the families who do not accept the continuation of a pregnancy with high risk of Down syndrome condition, since the abortion legal age is 12 weeks, in most countries (Grimes & Cates, 1978). This concern is present also in our country where 14 weeks, the age limit for abortion is. An exception of these legal limits is justified only in case of therapeutic purposes. Considering that Down syndrome does not threaten the life of the mother nor the child, ending of the pregnancy on this ground is not legally sustained. Also, termination of a late pregnancy is associated with higher distress for health-care professionals than ending an early pregnancy. The greatest concern is regarding the presence of life signs after abortion (Vinggaard & Petersson, 2012).

### ***Announcing the diagnosis of Trisomy 21***

It is fundamental to mention that this represents a very important and delicate moment. Exposure of the diagnosis before the family is a fundamental event for the future development of the child and family. The announcement marks a sudden rupture between what the child is in reality and what the parents expected. The announcement brings a shade of sadness of the parents on the “ideal child”, and the projects whose object the child will be (Tobolcea, 2002; Zuckoff, 2002). The pregnant mothers and those who have children with Down syndrome should: (1) look for a doctor with experience in treating children with Down syndrome; (2) inform themselves about Down syndrome (it is indicated consulting book stores and libraries, the Internet, in order to learn more about the Down syndrome); (3) participate in meetings of support groups (associations of children with Down syndrome).

Communication with other parents who have children with Down syndrome can also be helpful when dealing with difficult feelings. It is also useful to know not only the possible problems that may arise, but also to discover the joys that other parents had with their children. The parents of children with Down syndrome have an important role in helping the child to achieve his full potential. Moreover, to be the parent of a child with Down syndrome, involves a lot of challenges and frustrations and frequent changes of the emotional state - which can all lead to exhaustion. It is recommended that parents look after themselves, in such a way as to have the power to be with the child and to meet his needs. Also, all the parents with children having Down syndrome should group in associations to advocate for a better life for their children and to fight for their rights. It is important to know that children with Down syndrome are very receptive to what

is happening around them, as they are very affected when violence or harsh and strong words are used, which is why we recommend their families to do everything in their power to keep an atmosphere of peace and quiet in their home. These children need a lot of love and must have a good knowledge about love, as they respond to love in the same way (Tobolcea, 2002; Pueschel *et al.*, 1997).

## Conclusions

The screening test performed after 14-15 weeks it is useful in calculating the probability for Down syndrome and not for establishing the diagnostic and, further one has only an informative role without leaving the family to decide upon the development (or non-development) of the pregnancy. The interruption of pregnancy after 12 weeks is called therapeutic abortion and may be performed only due to precise medical causes. The Down syndrome benefits legal support and is not included in the categories of conditions that allow interruption of pregnancy through therapeutic abortion. However, increasing the access to social assistance and improving the management of the Down syndrome with respect to psychosocial and medical support is the most important goal.

Also, development of future non-invasive investigations, like MaterniT21, affordable and with high reliability, can give the possibility to the families facing such problems during pregnancy with physical, social and economical implications, to decide freely on their future destiny. In this way, further research in this area seems warranted and it is greatly encouraged.

## References

- Ahmed, S., Bryant, L.D., Ahmed, M., Jafri, H., & Raashid, Y. (2013). Experiences of parents with a child with Down syndrome in Pakistan and their views on termination of pregnancy. *Journal of Community Genetics, 4*(1), 107-114.
- Bjorklund, U., Marsk, A., Levin, C., & Ohman, S.G. (2012). Audiovisual information affects informed choice and experience of information in antenatal Down syndromescreening—a randomized controlled trial. *Patient Education and Counseling, 86*(3), 390-395.
- Capone, G.T., Aidikoff, J.M., Taylor, K., & Rykiel, N. (2013). Adolescents and young adults with Down syndrome presenting to a medical clinic with depression: comorbidobstructive sleep apnea. *American Journal Medicine Genetics, 161*(9), 2188-2196.
- Deborah, A., Driscoll, S.G. (2009). Prenatal Screening for Aneuploidy. *New England Journal Medicine, 360*, 2556-2562.
- Down's Syndrome Research Foundation (2005). *Bright Beginnings: A Guide for New Parents*. Buckinghamshire, UK: Down's Syndrome Research Foundation.

- Dykens, E.M. (2007). Psychiatric and behavioral disorders in persons with Down syndrome. *Mental retardation and developmental disabilities research reviews*, 13(3), 272-278.
- Geok, C.K., Abdullah, K.L., & Kee, L.H. (2013). Quality of life among Malaysian mothers with achild with Down syndrome. *International Journal Nursing Practice*, 19(4), 381-389.
- Grimes, D.A., & Cates, W.Jr. (1978). Gestational age limit of twelve weeks for abortion by curettage. *American Journal Obstetrics Gynecology*, 132(2), 207-210.
- Gronowski, M. (2004). *Prenatal Screening for Fetal Defects. In Handbook of Clinical Laboratory Testing During Pregnancy*. Totowa, NJ: Humana Press.
- Hattori, M., Fujiyama, A., & Taylor, T.D. (2000). The DNA sequence of human chromosome 21. *Nature*, 405 (6784), 311-319.
- Jones, E.L., Mok, K., Hanney, M., Harold, D., Sims, R., Williams, J., & Ballard, C. (2013). Evidence that PICALM affects age at onset of Alzheimer's dementia in Down syndrome. *Neurobiol Aging*, 34(10), 1-5.
- Kent, L., Evans, J., Paul, M., & Sharp, M. (1999). Comorbidity of autistic spectrum disorders in children with Down syndrome. *Developmental Medicine & Child Neurology*, 41(3), 153-158.
- Malt, E.A., Dahl, R.C., Haugsand, T.M., Ulvestad, I.H., Emilsen, N.M., Hansen, B., Cardenas, Y.E., Skold, R.O., Thorsen, A.T., & Davidsen, E.M. (2013). Health and disease in adults with Down syndrome. *Tidsskr Nor Laegeforen*, 133(3), 290-294.
- Mansfield, C., Hopfer, S., & Marteau, T.M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter Syndromes: a systematic literature review. European Concerted Action: DADA (Decision-making After the Diagnosis of a fetal Abnormality). *Prenat Diagnostic*, 19(9), 808-812.
- Natoli, J.L., Ackerman, D.L., McDermott, S., & Edwards, J.G. (2012). Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995-2011). *Prenat Diagnostic*, 32(2), 142-153.
- Nicham, R., Weitzdorfer, R., Hauser, E., Freidl, M., Schubert, M., Wurst, E., Lubec, G., Seidl, R. (2003). Spectrum of cognitive, behavioural and emotional problems in children and young adults with Down syndrome. *Journal of Neural Transmission, Suppl 1*, 1-5.
- Patrick, K., Wade, R., Goulden, N., Rowntree, C., Hough, R., Moorman, A.V., Mitchell, C.D., & Vora, A. (2014). Outcome of Down syndrome associated acute lymphoblastic leukaemia treated on a contemporary protocol. *Br Journal Haematol*, 165(4), 552-555.
- Pueschel, S.M., & Sustrova, M. (eds.). (1997). *Adolescents with Down Syndrome: Toward a More Fulfilling Life*. Baltimore, MD: Brookes.
- Tobolcea, I. (2002). *Dezvoltarea abilităților de limbaj și de comunicare la copilul cu sindrom Down (trizomia 21)*, Iasi: Spanda.
- Van Dyke, D.C.; Mattheis, P.J., Schoon, E.S, & Williams, J. (1995). *Medical and Surgical Care for Children with Down Syndrome*. Bethesda, MD: Woodbine House.
- van Schrojenstein Lantman - de Valk, H.M., Haveman, M.J., & Crebolder, H.F. (1996). Comorbidity in people with Down's syndrome: a criteria-based analysis. *Journal of Intellectual Disability Research*, 40(5), 385-399.



Vinggaard, C.A., & Petersson, B.H. (2012). Different attitudes among health-care professionals to late termination of pregnancy - a systematic review. *Ugeskr Laeger*, 174(16), 1084-1088.

Zuckoff, M. (2002). *Choosing Naia: A Family's Journey*. New York: Beacon Press.