The importance of cytogenetic screening in prenatal diagnosis of Down syndrome

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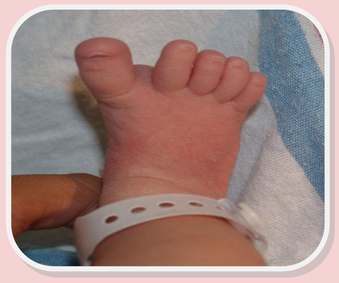
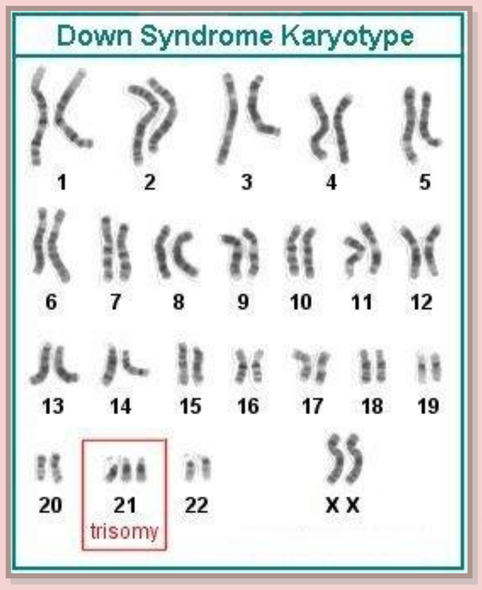
Introduction

Down syndrome, or trisomy 21 is a complex genetic disease resulting from the presence of 3 copies of chromosome 21.The origin of the extra chromosome is maternal in 95% of cases and is due to the failure of normal chromosomal segregation during meiosis.Early detection and diagnosis of DS are very important.Ages of parents and their choice whether to continue pregnancy or not should be respected.Most children with Down syndrome are born to mothers over 30 years of age.

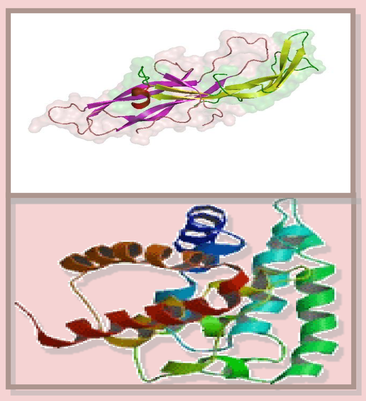
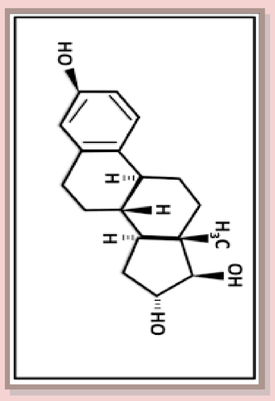


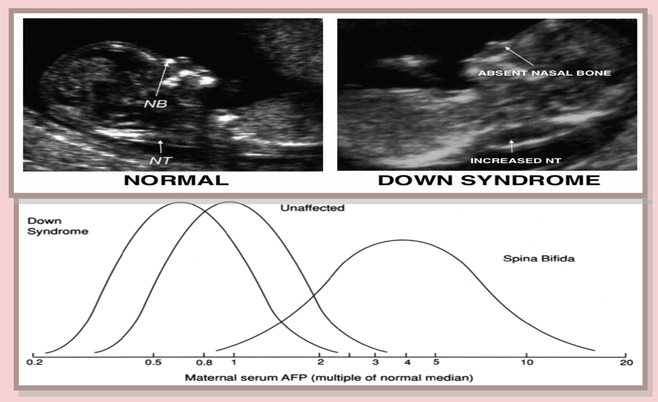




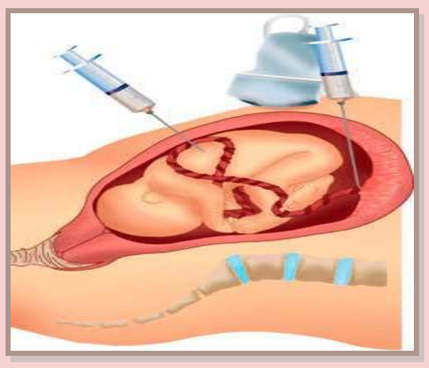
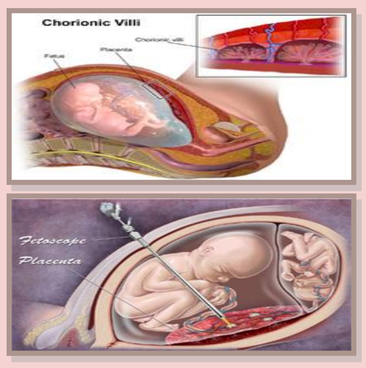

Material and methods

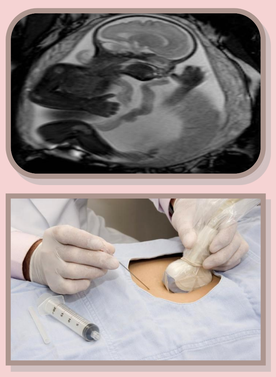
There are many cytogenetics methods for early detection and diagnosis for the DS, some of them are invasive and the others are non-invasive methods.

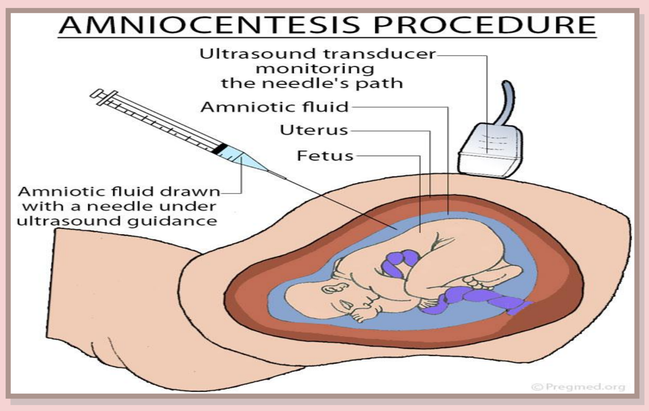
Non-invasive methods should be applied to every pregnant woman, while invasive diagnostics is indicated only in certain situations, such as women over 35god,the parent carriers of a genetic disease,family history of neural tube defect,parents who have previously had a child with genetic malformations,stillbirths or chromosome disorders of unknown cause,women with a positive triple test(abnormal level of serum alpha fetoprotein-AFP,human chorionic gonadotropin-HCG,unconjugated estriol-UE3),women exposed to radiation or infection,other maternal diseases(diabetes, epilepsy).



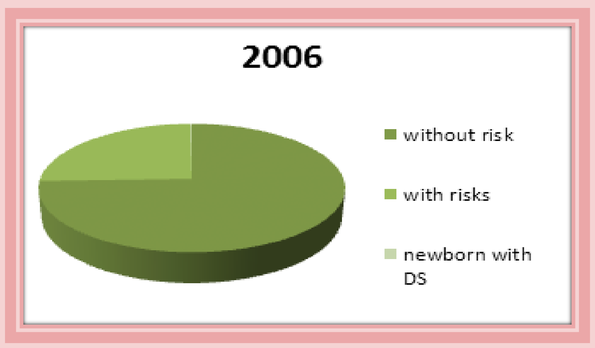



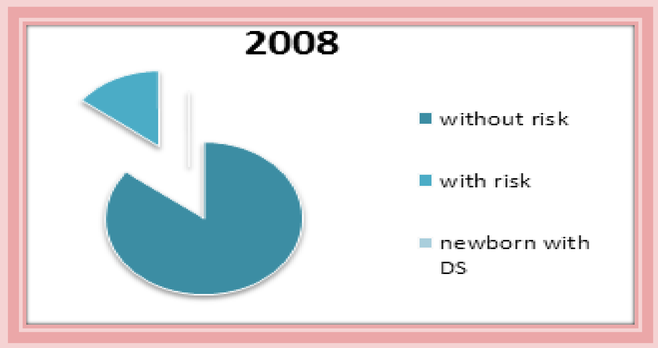




Results and discussion

In a period of ten years from 2006 to 2015 in city of Bitola there were born three babies with Down syndrome,all of them are baby girls.First two babies were born in 2006.As a risk factor for the first baby is considered the presence of the maternal disease-Diabetes.In 2008 as a newborn risk factor for DS was the age of the mother during pregnancy that amounted 39years.In 2006 there were 1048 newborns,782 of them were risk-free and 266 with risks for getting DS.In 2008 the total number of newborns was 1071,916 were risk-free and 155 with risks for DS.





Conclusion

Cytogenetic screening methods are essential,especially noninvasive which are made easy for every pregnant woman. This methods may discover genetic malformations of the fetus in the early pregnancy.Because recently screening showed presence of Down syndrome in younger mothers than 30 years it is considered as necessary.Expectancy and quality of life are much better nowadays thanks to modern medicine and medical techniques.