

Chapter 10

Automatic Computerized Diagnostic Tool for Down Syndrome Detection in Fetus

Michael Dinesh Simon
Anna University, India

Kavitha A. R.
Anna University, India

ABSTRACT

Down syndrome is a genetic disorder and the chromosome abnormality observed in humans that can cause physical and mental abnormalities. It can never be cured or rectified. Instead it has to be identified in the fetus and prevented from being born. Many ultrasonographic markers like nuchal fold, nasal bone hypoplasia, femur length, and EIF are considered to be the symptoms of Down syndrome in the fetus. This chapter deals with the creation of automatic and computerized diagnostic tool for Down syndrome detection based on EIF. The proposed system consists of two phases: 1) training phase and 2) testing phase. In training phase, the fetal images with EIF and Down syndrome is analyzed and characteristics of EIF are collected. In testing phase, detection of Down syndrome is performed on the fetal image with EIF based on the knowledge cluster obtained using ESOM. The performance of the proposed system is analyzed in terms of sensitivity, accuracy, and specificity.

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INTRODUCTION

Down syndrome is the chromosomal abnormality caused in humans when extra genes from chromosome 21 are transferred to a newly produced embryo. It causes a change in the fetal development leading to physical and mental abnormalities. The babies with Down syndrome have a distinguished appearance than normal babies. The child suffering from Down syndrome are likely to have retardation in growth and the mental problems. Down syndrome in general cannot be cured; it can be prevented by discontinuing the pregnancy based on the symptoms observed in the fetus.

There exist multiple ultrasonographic markers like nuchal fold, nasal bone hypoplasia, femur length that can lead to Down syndrome child. Recently, Echogenic Intracardiac Foci (EIF) had been added as a new observation in this line. Many clinical researches prove that presence of EIF acts as a possible symptom of Down syndrome. An EIF is defined as a bright white shape that is spotted in the heart of the fetus and it can be clearly visualised in an ultrasound image. An article “Ultrasound findings” reports that EIF is seen in about 1 out of every 20 or 30 pregnancies (~3-5%). If an EIF is observed in an ultrasound image, the mothers carrying the fetus with EIF undergo terrible mental pressure fearing that they would deliver Down syndrome child. This mental pressure created in the pregnant women can have undesirable impact on the fetus. To bring the down the mental pressure of the pregnant women and to ensure the fetus is unaffected with Down syndrome, women with EIF fetuses are suggested to undergo Chorionic villus sampling and amniocentesis.

Amniocentesis and Chorionic villus sampling methods are well known non-ultrasonic method for detecting Down syndrome. Amniocentesis is a process of collecting small quantity of the amniotic fluid that encircle the fetus and analyzing it for trisomy 21. It is performed with the help of ultrasonic guidance. It has disadvantage such as leaking amniotic fluid, miscarriage, needle injury to fetus and infection transmission. Chorionic villus sampling is a process of performing chromosomal analysis of cells taken from the placenta. It suffers from miscarriage and uterine infection problems.

Down syndrome is no more a rare phenomena; the affected rate is gradually improving throughout the world. A statistical report show that i) the risk of Down syndrome in the overall birth rate is 0.00125% ii) women in the age group of 30 and below have 0.001% risk of Down syndrome iii) women in the age group of 45 have 0.022% risk of Down syndrome. In spite of Down syndrome affected rate is being high, there doesn't exist an easy diagnostic method for its detection. This acts as the driving force for this research.

The major contributions of the chapter are i) a new medical parameter EIF is used for the detection of Down syndrome ii) extended region growing (ERG) is utilised

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