A new Non-Invasive Prenatal Diagnosis for Down Syndrome

Maternal DNA blood test during pregnancy can provide a safer and more effective prenatal diagnosis for Down syndrome

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I would like to extend a warm welcome to you all, and especially to the Members of the Media.

Please allow me, as the coordinator of this research, to thank all the scientists of my team, and especially Elisavet Papageorgiou and Evdokia Tsaliki for the hard work and dedication they have put into this research over the last 5 years. Without their efforts, the scientific achievement that we are presenting here today would not have been possible

I would also like to extend my thanks to our collaborators who worked with us on this research: Dr Alex Karagrigoriou, Associate Professor in the Department of Mathematics and Statistics of the University of Cyprus who developed the statistical analysis; Dr Voula Velissariou, Head of the Department of Cytogenetics and Molecular Biology at Mitera Hospital in Athens who collected the samples that were tested; and Dr Nigel Carter, leader of the Molecular Cytogenetics Team of the Wellcome Trust Sanger Institute in Cambridge, who assisted in the initial stages of the research for the development of epigenetic markers. My warm thanks to all.

Last but not least, I would like to thank the Board of Directors of the Cyprus Institute of Neurology and Genetics for their invaluable assistance to our scientists, and the Republic of Cyprus for its continuous support to the Institute. The Institute is a scientific center of excellence and as such gives us the opportunity to develop, here in Cyprus, pioneering research of an international standard. My thanks also goes to all

those who fund our research and especially the major supporter of this project which was the European Union through the 6th Framework Program.

I am very pleased to be able to announce today the development and validation of a novel test for non-invasive prenatal diagnosis for Down Syndrome.

Down Syndrome or Trisomy 21 is the most common cause of mental retardation with an incidence of 1 in 600 births. It is caused by the presence of an extra chromosome 21 which leads to physical and intellectual impairments.

The invasive procedures currently used for prenatal diagnosis of Down Syndrome, such as chorionic villus sampling (CVS) (in the 11-12th week of gestation) or amniocentesis (in the 16th week of gestation), involve the use of a needle which passes through the womb and collects placental cells or some of the fluid that bathes the fetus. Both procedures are associated with a considerable risk of fetal loss which is estimated to be 1% of the pregnancies. For this reason, today the prenatal diagnosis of Down Syndrome is offered only to pregnant women who have a high risk of giving birth to a child with Down Syndrome. These high risk pregnancies are identified with ultrasound and biochemical analysis and make up 8-10% of total pregnancies. Following the genetic investigation of high risk pregnancies, approx. 80% of the Down Syndrome are diagnosed. For these reasons, over the last few years, scientists have been looking for a new, non-invasive method of prenatal diagnosis which will be offered to all pregnancies, is not associated with any risk of miscarriage and will provide prevention to all incidents of Down Syndrome.

Dr Philippos Patsalis and his team have developed a new non-invasive prenatal test for Down syndrome which only uses a small amount of blood from the pregnant women. The study involved the collection of 10ml of blood, at 11-14th week of gestation, from 80 pregnant women, of which 34 were from pregnancies carrying a fetus with Down syndrome and 46 pregnancies carrying a fetus with normal constitution. All normal and all Down syndrome cases were correctly identified providing 100% specificity and 100% sensitivity. The development and validation of the novel method has just been published in Nature Medicine, one of the World's top ranking and most prestigious scientific journals.

The success of the test is based on the investigation of epigenetic markers on chromosome 21 which are found exclusively in the fetal DNA. In cases where the fetus has Down syndrome, together with the extra chromosome 21 there will be an extra copy of the fetal specific epigenetic marker. The test is able to quantify this extra copy of chromosome 21 and distinguish normal pregnancies from pregnancies bearing a Down syndrome fetus.

The new non-invasive test provides considerable advantages as compared to the current invasive procedures and therefore it is expected to be introduced with high confidence as a routine test in the clinical practice in the near future. The main advantages are: firstly, as a non-invasive test bearing no risks for fetal loss; Secondly, it can be offered to all pregnant women; and thirdly, it can provide effective prevention of Down Syndrome. The new test also provides the following advantages: provides 100% accuracy in the non-invasive prenatal diagnosis of Down Syndrome; the application of the new test is relatively simple as it does not require specialized or complex laboratory equipment, software, infrastructure or knowhow. Therefore, the test can be easily introduced into every genetic diagnostic lab in the world. The cost of performing this test is affordable and even cheaper than the current methods used for invasive prenatal diagnosis. Finally, it is also a fast method as the results can be obtained within 4-5 days.

Dr Philippos Patsalis and his team are now in the process of performing a larger-scale clinical study which is essential in order to enable the introduction of the new test into clinical practice. It is estimated that the new test will be introduced into clinical practice within the next two years. The team of Dr Philippos Patsalis is also researching the modification of the new test so it can be utilized for the non-invasive prenatal diagnosis of other syndromes.

The scientific publication can be found on the Website of *Nature Medicine:* http://dx.doi.org/10.1038/nm.2312

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