

Case Report

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## False Positive also Exists: Keep it in Mind when Dual Test is Performed



Piazze Juan<sup>1</sup>, D'Orazi Valerio<sup>2</sup>, Romanzi Daniela<sup>4</sup>, Rosselli Michele<sup>5</sup>, Bratta Massimo<sup>6</sup> and Silvio Rea<sup>3</sup>

<sup>1</sup>ASL Frosinone (Poliambulatorio di Ceprano-Ospedale SS Trinità di Sora), Fondazione Carlo Ferri Monterotondo, Italy

<sup>2</sup>Department of General Microsurgery and Hand Surgery, "Fabia Mater" Hospital, Italy

<sup>3</sup>Chirurgia Oncologica, Università degli Studi de L'Aquila, Italy

<sup>4</sup>Fondazione "Carlo Ferri", Monterotondo, Italy

<sup>5</sup>Unità operativa di Oncologia, Ospedale Civile di Frascati, Italy

<sup>6</sup>Ospedale Civile di Rieti, Fondazione Carlo Ferri Monterotondo, Italy

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**\*Corresponding author:** Juan Piazze, ASL Frosinone, Fondazione Carlo Ferri Monterotondo, Rome, Italy, Email: [cloudpia@me.com](mailto:cloudpia@me.com)

### Abstract

We report two cases of false positive results regarding Dual test for first trimester non invasive diagnosis of chromosomopaties, describing how different paths may be present even with simile parameters, in two different pregnant women.

**Keywords:** Dual test; False positive test; Ultrasound

### Introduction

The Down syndrome is a congenital disorder which verifies when a trisomy of 21th chromosome is present (T21). In Italy, 1 neonate over 700-1200 is born under this condition, which presents a variable degree of retardation in mental and physical development, other than the presence of other congenital malformations [1].

The prenatal diagnosis includes the analysis of fetal cells caryotype (blood or amniotic fluid) or from throphoblast (villocentesis) and a non-invasive procedure, the Dual test, which is performed when CRL ranges between 45 to 84 mm within 11 to 13+6 wks gestation [2]. The Dual test is an association of fetal nuchal translucency (NT) plus the evaluation of maternal free-BhCG and PAPP-A (detection rate 85-90%, false positive rate 3-5%). This Dual test may be enhanced by the evaluation of other ultrasound parameters such as the nasal bone, the ductus venous and the tricuspid regurgitation. In Italy, and as suggested by the SIEOG, the prenatal screening must be proposed to every pregnant woman, independent form age at the moment of examination. This information may be offered not only by gynecologist but also by midwives and the Family Doctor, or even by means of written material. Regardless of the prenatal diagnosis chosen, if a high risk is found (considered

1/250), the pregnant woman must be informed regarding the possibility of an invasive prenatal diagnosis, which includes the villocentesis in the first trimester and the amniocentesis in the second trimester [3].

We report two cases of false positive results regarding Dual test for first trimester non invasive diagnosis of chromosomopaties, describing how different paths may be present even with simile parameters, in two different pregnant women.

### Cases

Two pregnant women were referred to our department for a first trimester evaluation and for the strategy regarding the pregnancy follow-up. We underline the similarity in both women, one 33 (lawyer) and the other 32 year old (school teacher), both in second pregnancy with the first baby born under spontaneous delivery and in good neonatal conditions.

In both cases the Dual test was proposed and performed within the twelfth week gestation as stated linked to an optimal period for this method execution. After the Dual test responses were ready, both pregnant women presented the same peculiarities: nuchal translucency at 95° percentile and both maternal serum parameters altered. If just NT were altered, an

evaluation of maternal blood free placental DNA (cfDNA) would be proposed [4], if both parameters (NT and maternal serum) were altered, an invasive prenatal diagnosis procedure would be proposed. So the gynecologist and both the pregnant women decided to perform the amniocentesis between 16-18 wks gestation. And this is where this presentation changes, in one of the cases, every doubt was solved serenely, and the pregnant woman tried to solve every doubt or eventual risk directly with the gynecologist help.

In the other case, the 32 year old pregnant woman looked for other physicians suggestions (not gynecologist colleagues), and was constantly in contact with other women with amniocentesis recommended after altered Dual test, in the deleterious internet forums. But what the other pregnant woman absolutely did not do: the 32 year old woman made take a look of the Dual test response to a statistician!

When we observed these two ways of facing the same problem in two healthy women, we decided to face the truth and underline the only argument that may have tranquilized both of them, the false positive rate of about 3-5% regarding Dual test procedure. The 33 year old took a breath in her preoccupation, and the 32 year old went deep into the fact: "but are you sure, is it more a 3% or a 5%?"

What followed was absolutely the same in both cases, amniocentesis performed uneventfully, two normal caryotype fetuses in which a structural ultrasound examination by middle gestation and an echocardiography were performed showing two pregnancies in absolute maternal-fetal health conditions; both of them now by 32-34 wks gestation.

## The final analysis

The lawyer with a permanent laugh, the other lady facing the physicians group telling us: if you knew that false positive exists, why did you make me perform the Dual test!

## Comment

This more than a serious case-report, is a report of our clinical experience regarding the behavior of two women with the same risks, in simile conditions; that may not be absolutely the same.

When the 32 year old school teacher talked about the statistician that reviewed the Dual test response, we understood the female's world is certainly a wonderful unknown universe. And a pile of clinical practice reflections, women to whom an altered test is explained, are not acquainted with new terms (i.e. diagnostic accuracy, false positive, detection rate, multiple of median, sensibility, etc.). Some of them may accept what we try to explain, the 32 year old pregnant woman is an example that this may not always happen and that, at the end, she did everything in order to convince herself that her baby was a healthy one, fruit of that blessed false positive rate.

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