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Dizygotic Twins with Down Syndrome Complicating Pregnancy - A Unique Case

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Abstract

Scientific evaluation of current bibliography depicts the significance of prenatal diagnosis in terms of proper gestations surveillance.

Appearance of such chromosomal anomalies is strongly accompanied with mother age, previous obstetrical or gynecologic history, previous surgical procedures.

Ultrasound depiction of Nuchal Translucency (NT) reflects the tendency of such gestations to develop such anomalies, influencing further pregnancy course.

Final diagnosis, in cases of increased NT, consists Chorionic Villus Sampling (CVS) and further karyotype evaluation.

Among prenatal appearance of chromosomal anomalies, Trisomy 21 or Down syndrome represents the most common type with characteristic morphologic and clinical diagnosis.

Prenatal diagnosis of dizygotic twins of opposite sex type with simultaneous appearance of Down Syndrome is very rare throughout current literature.

Our study reflects presentation of such unique and rare case with proper diagnosis and treatment.

Keywords: *Trisomy 21, Down Syndrome, Nuchal Translucency, Chorionic Villus Sampling.*

Introduction

Multiple gestations, are increased especially in women with infertility issues as IVF (In Vitro Fertilization) procedure.

Maternal age is strongly accompanied with development of chromosomal anomalies with most characteristic representative the prenatal diagnosis of Trisomy 21 or Down Syndrome(DS). [1]

During prenatal screening measurement of NT(Nuchal Translucency) reflects proper diagnosis of such cases.

Along with maternal age, presence of nasal bone, serum ranges of PAPP-A (Pregnancy Associated Plasma Protein-A) and free Beta Chorionic Gonadotropin (B-HGD), it can be assumed statistically the possibility of chromosomal anomalies establishment.

According to current bibliography, it is very rare for both membranes of a twin pair to be concordant for DS.

Dizygotic twins with Down Syndrome represent a very rare entity estimated about 4 per 10 million births. [2]

In cases with increased NT, CVS (Chorionic Villus Sampling) depicts next step of prenatal screening.

Karyotype analysis will confirm and establish final result excluding positive or negative false results.

Genetic counselling is mandatory in order to guide the couple deciding the appropriate solution.

In such cases with prenatal diagnosis of dizygotic twin pregnancy with DS, therapeutic mapping depends on final decision of the couple.

Objective of our study consists of presentation of such unique case, informing all scientific community of proper diagnosis and optimal treatment.

Case

We present a case of 40-year-old female patient (gravida1, para1) with negative gynecologic history admitted to our Department with signs of vaginal bleeding and episodes of vomiting.

Among assiduous laboratory examination, free Beta Chorionic Gonadotropin (B-HGD) in serum revealed ranges of gestation.

Transvaginal ultrasound evaluation, confirmed presence of intrauterine and multiple gestation simultaneously.

Twin pregnancy as result of natural conception was uneventful, not revealing severe complications during surveillance. (Blut type AB Rhesus positive, VDRL negative, HbS negative, HbsAg negative,HIV control negative, TORCH (Toxoplasma, Rubella, Cytomegalovirus, Herpes) negative, antiHCV negative, Hct 34,8mg/dl /Hb11,4mg/dl, WBC 7300 mg/dl,PLT 262,000mg/dl, Gu 95 mg/dl.

At 12²weeks of gestation, prenatal screening measurement of NT(Nuchal Translucency) was performed for both fetuses.

Fetus A: *Positive Heart Frequency, NT 2,50 mm/BPD (Biparietal Diameter) 24,0mm/HC (HeadCircumference) 80,0mm/AC(AbdominalCircumference)70,0mm/FL (Femur Length) 7, 1mm/ Amniotic fluid normal and nasal bone present.*

Fetus B: *Positive Heart Frequency, NT 2,80mm/ BPD (Biparietal Diameter) 22, 0mm/HC (Head Circumference) 80,0mm/AC (Abdominal Circumference) 71, 0mm/Amniotic fluid normal and nasal bone present.*

Endocervical length measured 40mm. These ultrasound imaging findings, consisted statistical possibility of Trisomy 21 or Down Syndrome 1/46 for Fetus A and 1/20 for Fetus B respectively.

With the addition of free Beta Chorionic Gonadotropin (B-HGD) and PAPP-A (Pregnancy Associated Plasma Protein-A) *final statistical possibility for Trisomy 21 or Down Syndrome increased in* ***1/32 for Fetus A*** *and* ***1/14 for Fetus B*** *respectively.*

Facing this reality, next step of our therapeutic mapping consists of Chorionic Villus Sampling (CVS) and further karyotype evaluation. Two individual longterm cultures of chorionicvilli have been setup and a total of 20 meta phases have been studied, after the application of GTG(Giemsa)-banding.

Banding of chromosome with enzymes and stains is essential to identifying normal and abnormal chromosome structures.

Cytogenetic analysis revealed that all studied metaphases contain 47 chromosomes, with presence of an extra copy of chromosome 21 (trisomy 21) both of two fetuses. **Female Fetus A (*Figure Ia.) and Male FetusB (Figure Ib.)*.**

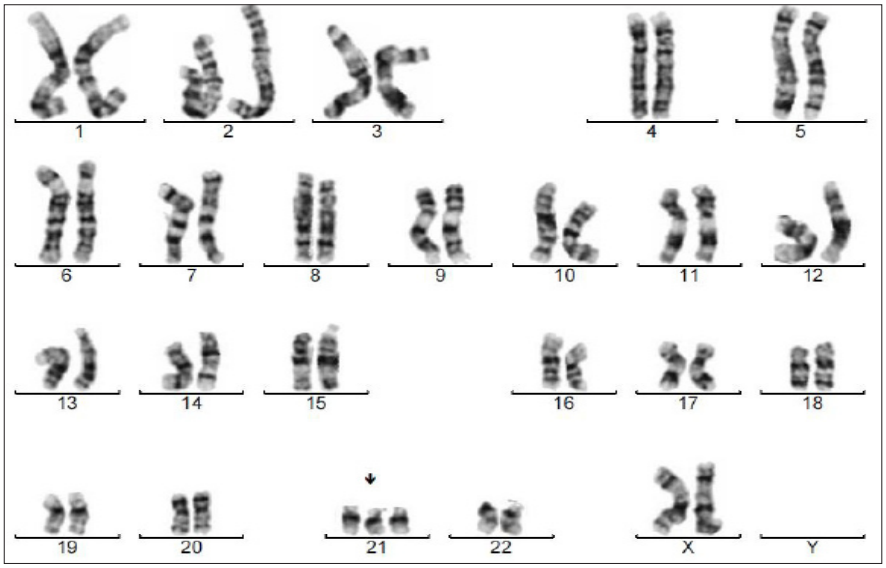

Fig Ia. *Karyotype of a female with trisomy 21.*

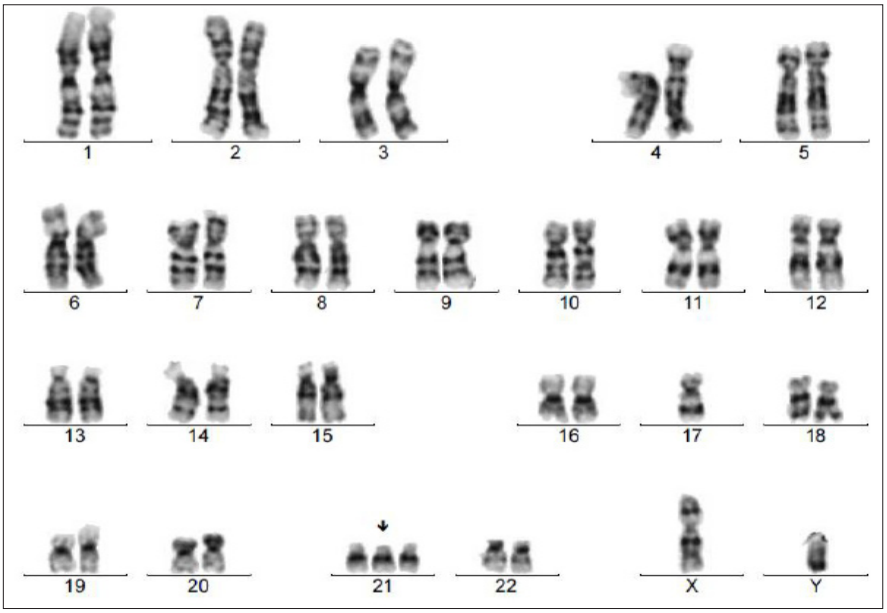

Fig Ib. *Karyotype of a male with trisomy 21.*

No other structural or numerical chromosomal abnormalities were observed. In 15th week of gestation, therapeutic evacuation of the uterus was performed following couple'sdecision. ***(Figure II.)*** Following opd, patient was discharged from hospital in good clinical condition.

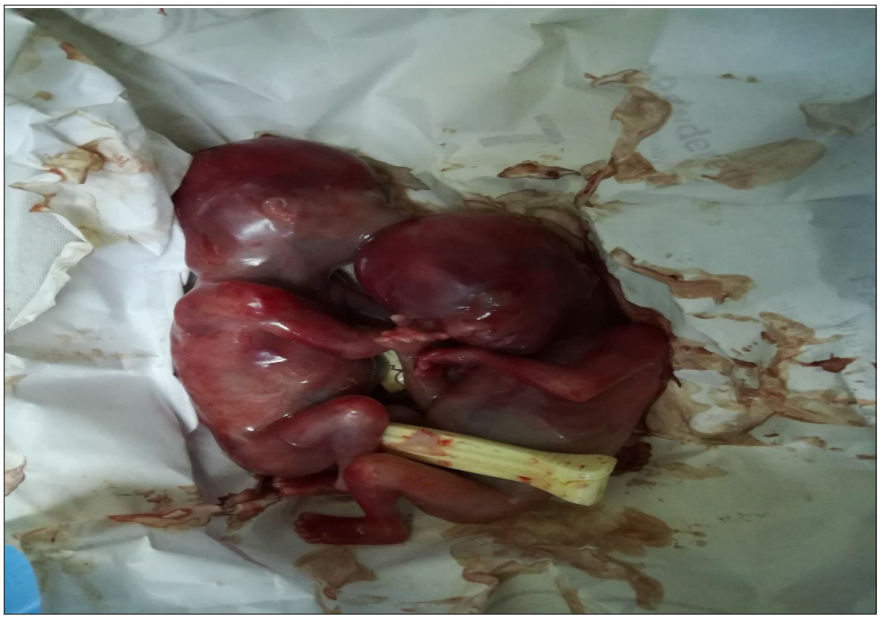

Fig II. *Dizygotic twins with Down Syndrome*

Discussion

Dizygotic twins as result of natural conception with fetuses of opposite sex type, both infected consist a very unique entity throughout current bibliography.

Birth defects (whether chromosomal or non-chromosomal) are common, costly and critical.

Incidence of Trisomy 21 or Down Syndrome is strongly associated with mother age. [3]

As mentioned above, it is very rare of a twin pair to be concordant for Down's Syndrome.

In such cases, reported incidence estimated lower than expected justifying as pathophysiologic mechanisms intrauterine death one of both membranes. [4]

Many hypotheses have been conducted in order to explain this rare entity. Increased argument reflects the mother age as in our case.

Many assumed the possibility of non-disjunction at the first meiotic division in the female, followed by fertilization of the egg nucleus by one sperm, and concomitantly of the second polar body by a different sex chromosome carrying sperm. [5]

Only two cases concordant for Down's syndrome have been reported in dizygotic twins of unlike sex.

Nicholson and Keay, first described a case of dizygotic twin pain of opposite sex without karyotype analysis, based only on clinical imaging findings. [6]

Fielding and Walker (1972) reported Down's syndrome in twins of opposite sex with simultaneously infection. They assumed that the incidence in such cases estimated less than 1:300,000.

Without doubt, such rare cases need genetic counselling, informing and explaining the couple the severity of the situation.

Therapeutic strategy will be based strictly on couple's decision. Mostly of cases therapeutic evacuation of the uterus consists gold standard.

After the therapeutic procedure, patient informed, due to her increased age, the possibilities of new fetal chromosomal establishment.

Aim of our study remains, presentation of such rare case, proper diagnosis and treatment.

Many studies must be conducted, in order to establish the correct pathophysiologic mechanism of such cases leading toultimate therapeutic mapping.

Conclusion

Dizygotic twins especially of opposite sex type concordant with Down Syndrome represents a very rare case unique in Greek current literature.

Aim of our study consists proper prenatal screening, assiduous diagnosis and treatment.

Even though in most of such cases, therapeutic evacuation of the uterus consists the gold standard, final therapeutic strategy depends on the decision of the couple.

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