

# A very rare genetic disorder presented with distinctive phenotypic features

## Abstract

**Background:** Mutation rates are not constant and are not limited to a single type of mutation; therefore there are many different types of mutations. Duplication is one of the genetic mutations. Diagnosis can be difficult since the number of chromosomes is normal.

**Case Report:** Patient 27 year old, G1P0. BMI 19. Non consanguineous parents, No alcohol user, non-smoker. No diagnosed genetic diseases in family. Ultrasound in 12w6d showed was normal. Normal NT, present nasal bone. Bi test showed low risk for trisomy 21,13,18. Ultrasound at 19 weeks found normal size fetus, normal AFI. At 21w6d fetus were observed the following features: Macrosomia fetal biometry for 25w1d, fat extremities and neck, macroglossia, hypospadias, thick nuchal fold. Polhydramnios AFI 29 cm. Amniocentesis was performed. Banded chariotype was performed. The pregnancy was interrupted. The fetus weight at 22 weeks was 945g. All the anomalies detected in the ultrasound were confirmed.

**Results:** 46 XY, arr 6q23.2 (131,643,624-132,146,514x3) 21q22.12 (36,926,448-37,439,348x3).

**Interpretation:** Male fetal profile with duplication in chromosome 6 in region 6q23.2 and duplication in chromosome 21 in the region 21q22.12.

**Discussion:** The fetus phenotype was very similar to Beckwith Weidmann syndrome. BWS results from various abnormalities affecting the proper expression of genes that control growth within a specific region of chromosome 11 (11p15.5). This region is referred to as the BWS critical region. Most common clinical features: Macroglossia, Exomphalos and gigantism in neonate, hemihyperplasia resulting in visceromegaly.

In our case the long arms of the chromosomes 6 and 21 were affected. A rare chromosomal disorder involving duplication of the long arm (q) of chromosome 6 which results in various abnormalities depending on the size and location of the portion of duplicated genetic material. Affects males to females 2:1. The fetus phenotype was very similar to the disorders involving the mutation of the chromosome 21.

**Keywords:** duplication, macrosomia, macroglossia

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## Case presentation

BN a 27year old G1P0 white female BMI 19, non consanguineous parents, no alcohol user, non-smoker. No diagnosed genetic diseases in family. Has done the prenatal tests that consisted in normal 12 weeks 6days ultrasound findings with NT 1.35mm, present nasal bone, low risk for trisomies 21,18,13. Normal ultrasound findings at 19weeks. But the morphological ultrasound at 21 weeks 6days showed a complex of abnormal ultrasound findings that are: Macrosomia fetal biometry for 25w1d, fat extremities and neck, macroglossia, hypospadias. Polihydramnios AFI 29cm. Amniocentesis was immediately performed. Distended panel was asked. The pregnancy was interrupted. The fetus weight at 22 weeks was 945g. All the anomalies detected in the ultrasound were confirmed.<sup>1-3</sup>

## Results of the amiocentesis

46 XY, arr 6q23.2 (131,643,624-132,146,514x3) 21q22.12 (36,926,448-37,439,348x3)

**Interpretation:** Male fetal profile with duplication in chromosome 6 in region 6q23.2 and duplication in chromosome 21 in the region 21q22.12 (Figure 1).



**Figure 1** Complex of abnormal ultrasound findings.

## Discussion

Since the fetus phenotype was very similar to Beckwith Wiedmann syndrome, we expected the results of amniocentesis to show chromosome 11 mutations. BWS results from various abnormalities affecting the proper expression of genes that control growth within a specific region of chromosome 11 (11p15.5). This region is referred to as the BWS critical region. Incidence 1:13700 live births, 85% of the cases are sporadic, 15% are familial. Most common clinical features: Macroglossia, Exomphalos and gigantism in neonate, Hemihyperplasia resulting in visceromegaly. But in our case chromosome 11 was normal

A rare chromosomal disorder involving duplication of the long arm (q) of chromosome 6 which results in various abnormalities

depending on the size and location of the portion of duplicated genetic material. Affects males to females 2:1. According to a study of 1990 Division of Genetics, Department of Pediatrics, University of Tennessee, USA, about duplication of chromosome 6 anomalies, It appears that duplication of the distal long arm of chromosome 6 (6q26-\*qter) produces a distinct phenotype. Patients with a phenotype which includes microcephaly, acrocephaly, downward slanting palpebral fissures, telecanthus, micrognathia, and carp shaped mouth, a characteristic short, anterior, webbed neck, club foot, joint contractures, and profound psychomotor retardation should be suspected of having a duplication of the distal long arm of chromosome 6. These inconsistent features may be attributed to the length of the duplicated segment of chromosome 6. But none of these features were observed in our case.

The duplication of 21q22.12, affects males to females 1:1 and the morphological features are very similar to trisomy 21.

Our case showed morphological features more similar to the mutations of the chromosome 21 than the mutations of chromosome 6. This might be due to the length and the site of the duplicated chromosome (Figure 2).



**Figure 2** Prenatal diagnosis of an abnormal fetus.

The ultrasound performed at 19 weeks showed no abnormalities. But only 3 weeks later at 21 weeks + 6 days ultrasound found completely different features. This fact suggests the morphological ultrasound might be better performed at 22 weeks than at 19 weeks.

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### Conflicts of interest

The author declares that they do not have any conflicts of interest.

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