

## Prenatal diagnosis in a mentally retarded woman with mosaic ring chromosome 18

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We present a pregnant woman with mental retardation and mosaic for ring 18 referred for prenatal diagnosis. Major clinical features included short stature with clinodactyly in feet, foot deformity and club feet, hypotonia, kyphosis, and absence of breast development, low set ears, high arched palate, dental decay and speech disorder. Prenatal diagnosis was carried. Using amniocentesis. The fetus had a normal karyotype described as 46,XX. The fetus was evaluated for clinical features after delivery; she was healthy with no abnormal clinical characterizations.

**Key words:** Mental retardation, prenatal diagnosis, ring (18)

### Introduction

The first case with ring (18) was reported in 1960s, more than 50 cases with r(18) have been presented since then. The clinical features for such patients are variable and depends on the extend of the deleted regions on the ends of chromosome 18. These patients usually have mental retardation, hypotonia, short stature, Microcephaly, developmental delay. Other characterizations which have been presented in cases with r(18) include heart defect, hearing loss, kidney problems, facial features, agammaglobulinaemia, insulin dependent diabetes mellitus. These features have also

been presented in patients with del(18q) and del(18p). Most patients with ring(18) share the clinical features with cases of del(18q).

To our knowledge this is the first patient with ring(18) evaluated for prenatal diagnosis where fetus had a normal female with no abnormal phenotypical characterizations

### Materials and Methods

An unrelated couple was referred for evaluation and genetic counseling for prenatal diagnosis. The woman was 22 years old. She had mild mental retardation (MR) with no family history of MR. Her husband was also severely mentally retarded with family history of mental retardation. He had a brother with MR with two normal sisters, his mother also suffered from mild mental retardation. The couple was the result of a consanguineous marriage. The husband's mental retardation was due to meningitis during newborn period according to parent's statement. The couple was married for 1 year and the wife was pregnant at 16 weeks of gestation when referred for prenatal diagnosis. The clinical features of the woman included short stature with clinodactyly in feet, foot deformity and club feet at birth and also dysplasia in nail, hypotonia, kyphosis and absence of breast development; facial abnormal features were low set ears, high arched palate, dental decay, and she had speech disorder, aggressive behavior and ataxic gait [Figure 1]. Chromosomal analysis using peripheral bloods for both couple was carried out. Chromosomal analysis was performed using standard GTG banding technique. The woman's karyotype

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Figure 1: (a) Mother with low set ears. (b) Short stature. (c) Club feet.



Figure 2: A normal female baby.

was mosaic for ring chromosome 18, described as 46,XX,r(18)(p11.3q23)[14]/46,XX[36] and the husband's karyotype was 46,XY. Amniocentesis was carried out in order to investigate the fetus's chromosome for ring18 and other possible abnormalities. The amniocentesis result demonstrated that the fetus had a normal female chromosome complement described as 46, XX. The fetus was followed up after delivery for evaluation and clinical characterizations. The baby was healthy with no abnormal clinical features [Figure 2].

### Discussion

The patients with ring 18 usually have similar phenotypic features to 18p deletion syndrome<sup>[1]</sup> or 18q deletion syndrome<sup>[2]</sup> or a combination of both features<sup>[3]</sup> including mental retardation, hypotonia, short stature, microcephaly, proximal or anomalous thumbs, speech delay, facial anomalies<sup>[4,5]</sup> agammaglobulinaemia,<sup>[6]</sup> growth hormone deficiency,<sup>[7,8]</sup> and insulin-dependent diabetes mellitus.<sup>[9]</sup> The mother had most of the phenotypic characterizations of ring18 as reported previously. The reports on patients with mosaic for ring18 are too few, hence there is a need for reporting more patients with r(18) in order to determine the distinguishing features between mosaic and nonmosaic for r(18).

Mothers with primary or mosaicism for r(18) have been reported with children affected for r(18).<sup>[10,11]</sup> To our knowledge this is the first reported case ascertained due

to referral for prenatal diagnosis leading to the delivery of a normal baby.

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