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Postnatal mosaic trisomy 16 (mt16), A rare case in Karachi, Pakistan

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Abstracts
Prenatally diagnosed Trisomy 16 mosaicism is related with variable pregnancy outcomes accepted from stillbirth with multiple congenital abnormalities and it is common as well in mosaic trisomy 16, huge level of trisomy 16 is mismatched with postnatal existence with maximum trisomy 16 conceptions impulsively aborting within the gestation period of 8 to 15 weeks. It is still unclear whether the level of trisomy in dissimilar fetal-placental tissues is an interpreter of pregnancy outcome or not. Cytogenetic analysis was carried out with blood lymphocytes and the metaphases witnessed with mosaic trisomy of chromosome.

Keywords: mosaic trisomy 16, postnatal, congenital abnormlities, cytogenetic analysis

Introduction
In spontaneous abortuses, up to 30% of the autosomal trisomies observed as a trisomy 16 is most common [P. A. Jacobs, *et al*; T. Hassold, *et al*; J. Wolstenholme,; F. Petracchi, L *et al*] [13]. Mosaic Trisomy 16 diagnosed postnatally at amniocentesis has rarely been reported and is even more uncommonly seen in postnatal cases [J. Wolstenholme, P. Benn, *et al*; M. T. Su, *et al*] [13]. Trisomy 16 mosaicism which is diagnosed prenatally is communal and linked with variable pregnancy results reaching from stillbirth with multiple congenital anomalies to seemingly normal newborn, creating genetic counseling most challenging. It is unclear whether uni-parental disomy (UPD) 16 donates to the phenotype, though it has been recommended that maternal UPD 16 distresses at some level of intra-uterine growth retardation (IUGR) and congenital anomalies. In these type of cases, the clinical topographies and results were highly inconstant with no identifiable phenotype. On the other hand, cases of trisomy 16 mosaicism are routinely encountered prenatally. Most commonly observed trisomy is Trisomy 16 in spontaneous abortions distressing around 1% of all conceptions [Hassoldand Jacobs, 1984; Wolstenholme, 1995] [13]. In mosaic trisomy 16, a high level of trisomy 16 is incompatible with postnatal survival with most trisomy 16 conceptions spontaneously aborting between 8 and 15 weeks gestation. Only 2 specious exceptions have been stated as [0] enduring into second [Cusik *et al*., 1995] [11] or third trimester [Yancey *et al*., 1996] [12]. Since the miscellany of results in prenatally ascertained trisomy 16 mosaicism and the rareness of long-term follow-up data, counseling on genetic level is complex. When mosaic trisomy 16 is seen at CVS, it is frequently the result of restrained placental mosaicism (CPM). CPM for trisomy 16 is linked with placental [1] inefficiency and growth retardation [Wolstenholme, 1995] [13], though a diversity of anomalies have been stated, the long-term consequence in the majority of cases that are survived seems to be good [Langlois *et al*., 2006] [14]. Mosaic trisomy 16 identified at amniocentesis is associated with a higher risk of abnormal outcome, cases usually showing fetal-death-in-uteropreeclampsia, intra-uterine growth retardation (IUGR), neonatal death, preterm delivery, congenital heart defect, developmental delay and some other minor abnormalities [Benn, 1998]. However latest reports may propose that a substantial proportion of prenatally identified patients with mosaic trisomy 16 have slight phenotype changes but good outcome [Langlois *et al*., 2006] [14]. It is not clear whether the level of trisomy in different fetal-placental tissues is a predictor of pregnancy outcome [Hsu *et al*., 1998; Yong *et al*., 2003] [6, 17].

Case presentation
According to this case; 10 years old male patient have autism spectrum disorder and obese (height;161cm, weight; 87kg) and He suspected for the klienfelter syndrome due to the Bilateral gynaecomastia noticed at the age of 7.5 years and micro size penis was observed. His parents were consageneouse couple and has unremarkable family history. Paternal age at conception was 25years for the mother and 28 years for the father. Mother had not the history of miscarriage. The problems observed in their first child otherwise learning issues has examine in their second child. Patient delivery history was full term and with SVD. According to the case condition, further clinical diagnoses tests were performed. Thyroid function test and lipid profile test revealed the normal in range but the serum insulin was 66.90 ulU/ml (Reference range 2-25ulU/ml), serum FT4 was 0.88ng/dl s(Reference range 0.89-1.76ng/dl), FSH male was 0.22 (1.27-19.26mlU/ml), LH was 0.10 (1.24-8.62mlU/ml), Testosterone hormone was 7.1 (6-27mlU/ml). Otherwise, the Prader-willi syndrome revealed the negative results. Cytogenetic analysis was carried out with blood lymphocytes, the metaphases observed with mosaic trisomy of chromosome.

Discussion
The diagnosed with chromosomal anomaly sixteen condition that was confined to fibroblasts and discovered postnatally. In each cases, no trisomic cells were found in blood cells. The

patients have terribly totally different outcomes reflective the variability of mosaic body abnormalities. Excluding cases of chromosomal
anomaly sixteen condition delineate before the introduction of body band, there area unit solely 3 reports of chromosomal anomaly sixteen condition during which the identification was established postnatal. The 5 according cases illustrate the broad variability of outcome, starting from death to a gentle constitution with traditional development. nonetheless I UGR, abnormal pigmentation of skin, uneven craniofacial and body findings, deafness, abnormally, scoliosis, and inborn heart defect square measure all clinical options delineated on many occasions in mosaic chromosomal aberration sixteen diagnosed pre- and postnatally (Gilbertson *et al*., 1990; Greally *et al*., 1990, 1996; Simensen *et al*., 2003; Yong *et al*., 2003; Ousager *et al*., 2006). [18, 19, 17] Confirming the diagnosing of chromosomal aberration condition is useful for one or two because it confers an occasional repeat risk for any pregnancies, sixteen condition may conjointly cause study the result of those cases and facilitate the direction, that is presently complicated as a result of the dearth of information

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