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**Case Report**

# Is genetic diagnosis a bliss or a bane in indian society? An impact on nuclear family: a case report of foetal skeletal dysplasia

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## ABSTRACT

Genetic counseling has been becoming an integral part of reproductive medicine practice. In area of feto-maternal medicine findings of ultrasonography are very important. Genetic counselor tries to link dots between USG findings and their genetic base. If abnormality is found, after the legal termination of pregnancy, for clinicians and patients, main concern is about its recurrence in future pregnancies. But the question arises about the acceptability of genetic risk calculations by concerned patient and her family. As in society like ours, the genetically literate population is quite low and generally not well prepared to accept and understand the risks related to genetic abnormalities. This thing makes work of genetic counselors much difficult when the genetic aberration is found in the concerned couple. This raises a question about the social acceptance of genetically different individuals who are phenotypically normal but if they open up about their abnormal genetic status there will be a risk of decrease in their social acceptability. Hereby we present a case report which forced us to pause and think about the level of depth at which we as a society reached so far to accept genetic analysis as one of the diagnostic tool in routine clinical practice.

**Keywords:** Genetic counselling, Antenatal scan, Skeletal dysplasia, Genetic aberrations, Marital harmony

## INTRODUCTION

Antenatal ultrasonography (USG) to know aneuploidies and morphological structural anomalies has become a routine investigation. A detailed scan between 11-13 weeks is very widely unfolding many issues related to genetics/chromosomes.[1] The utility of knowing morphological syndromes has been further exploited to requisition certain genetic tests like that of foetal tissue, foetal blood, cell free fetal DNA to know the genetic linkage by FISH or Microarray. When evaluated in depth many more correlating aberrations though within normal range of acceptability can be visualized with 3D USG.

The experts of society of fetomaternal have foresight to look for many other anomalies of other organ systems and fit that anomaly into a syndrome which may be inherited further and thus constitute important point for genetic counselling. Anomaly can be related to structural aberration or functional change seen on 3D USG or Color Doppler use. Colour Doppler at that stage explains the impending events related to uteroplacental unit. Even Preeclampsia can be predicted in a patient with pulsatility index of uterine arteries.

Genetic Testing can be described as the exploration of human chromosomes, DNA, RNA, proteins and particular metabolites in order to reveal heritable disease related mutations, phenotypes, genotypes or karyotypes for use in clinical practice. The results of genetic test can help to confirm or preclude a suspected genetic condition. There are more than 1,000 genetic tests which are currently in use and more are being developed. The interpretation of the results of genetic tests helps to determine an individual's chance of developing or passing on a genetic disorder. Various methods have been employed for genetic testing like molecular gene tests which study single genes or short lengths of DNA to

identify variations/ mutations leading to genetic disorder. Other test is chromosomal genetic test to analyse whole chromosomes to look if there are large genetic changes which can be an extra or missing copy of chromosomes causing a genetic condition. The amount of activity level of proteins or any abnormality can suggest changes to DNA resulting in genetic disorder which can be studied by biochemical genetic tests.[2]

In current article we report a case that came with routine antenatal level II anomaly scan for second opinion and decision making, if termination of pregnancy would be a better choice for skeletal dysplasia. Skeletal dysplasia is mainly a bone and cartilage disorder causing dwarfism and also it includes overgrowth syndromes. A number of genetic mutations can impede the organization and function of growth plate.[3]

## CASE REPORT

An apprehensive well educated, higher middle class income group couple came in the outpatient section of tertiary healthcare hospital for midtrimester termination of foetus with skeletal dysplasia. The wife's age was 33yrs and husband was 34 years old. The USG findings showed single foetus with Skeletal Dysplasia at 18 weeks 5 days. All long bones of appendicular skeleton were short and broad. All lay below first percentile for gestational age, mineralization of skull and spine was seen, thorax was narrow, ribs were short, amniotic fluid normal. There was no facial dysmorphology. The elective termination of fetus was done and requisite tissue for genetic testing was sent (skin biopsy and foetal blood).

The foetus karyotyping results showed low mitotic index. The metaphases showed pulverized chromosomes. There was a loss of number of chromosomes. The constitution of the karyotype was 41,Y,-X,-9,-11,-12, -22/46, XY.

To rule out the possibility of future child with any syndrome, karyotyping of couple was also done. The results of couple karyotyping came out to be a true shock. The result of female karyotyping came out to be 46,XX in 50% of metaphases scored, but there was loss of chromosomes in number of metaphases. The results showed approximately 20% metaphases with numerical anomalies, there were monosomies of chromosome 11, 20, 21 in 6% of metaphases. 14% of metaphases showed structural numerical anomalies, 6% of metaphases showed loss of chromosome 11, 18 and 4% of metaphases showed loss of chromosome 20, 21.

The karyotyping report of male showed 52% of scored metaphases with normal karyotype 46, XY. 33% of metaphases were having numerical abnormalities including loss of chromosome 16, 19, 20, 21, 22. Out of these metaphases with numerical abnormalities in 42% of metaphases chromosome 22 was missing. Around 15% of metaphases showed structural abnormalities including premature centromere division (PCD); an abnormal behavior of centromere which makes an individual susceptible to cell division errors due to chromosomal instability.

The results of couple predicted the probability of future with one or another genetic abnormality. Subsequently patient had ectopic pregnancy which was managed conservatively. The patient was under treatment for secondary infertility and showed significant concern related to genetic aberrations.

A year later couple went for a chromosomal reanalysis. The genomic instability in both of the individuals had increased. The wife showed 70% metaphases which were hypodiploid i.e. the number of chromosomes in a metaphases were between 37-43 chromosomes. No repetitive loss of chromosome was observed. Mosaicism was seen in 30% normal metaphases and 70% with numerical anomalies.

The male report showed 47% of aberrant metaphases. 13% of metaphases showed structural anomalies with chromosome breaks and chromosome gaps. 23% of metaphases showed numerical anomalies with deletion of chromosomes 5, 8, 10, 20, 22. Mosaicism was being observed in 47% metaphases showing non clonal chromosomal aberrations and 53% normal metaphases.

## DISCUSSION

The couple was explained the risk relationship of these chromosomal anomalies with respect to subsequent conceptions. The interpretation of the tests was desired by the couple in simple words related to each prospective issue. The reports showed genomic instability where the true attribution to causation in foetus is still worth analysis. To a clinician and concerned couple it simply matters if it occurs in subsequent pregnancy or not.[4] So it was apprised to them in well meaning words keeping in mind their receptivity through their expressions and reactions.

A follow up visit revealed that the outcome of each anomalous sequence and future concerns was not easy to be accepted by couple. The prognosis of future pregnancies and long term consequences were stressors for the couple. This created a near marital disharmony and non acceptability of the future issues creating a family dispute. The burden of disease was not shared but single handedly the female bore the brunt. The feeling of guilt was aroused in the female and no family support was provided.

The lust to conceive again was lost. Their reluctance for further investigations or genetic support was expressed by them. The wedlock commitments seemed to be crashing down. Genetic counselling for a brighter prospect was again explained and a precarious situation of genetic counselling was experienced making us realize

whether genetic diagnosis is a bliss or a bane in Indian society.

## CONCLUSION

The question arises: Are we in the social setup ready to accept the consequences of genetic counselling? Since we know every calculation has a pitfall and negativity, can it be turned into a positivity and constructive counselling to keep the couple and their marital harmony intact and also give them a child of their own which is a bliss by nature challenged by the geneticist? Can we mastermind and handle the genetics by use of advancing techniques or ask for genetically proven normal embryos be used in these couples or by surrogacy have a genetically normal healthy child and maintain marital harmony which is much desired in Indian society?

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