Abstract ID: - 213

Abstract Topic: - Clinical Genetics

Abstract Title: - Microarray and Karyotyping synergistically contributing towards clinical reporting

Presenting author name: - Jaydeep Akbari

Presenting author institute: - Genexplore Diagnostics and Research Centre Pvt. Ltd.

Co-authors name: - Bhumika Vaishnav, Jitendra Nayak, Vidhi Sambhvani, Jinal Parmar, Arpitsen Parmar, Alpesh Patel, Shivashankaran Chettiar, Devendrasinh Jhala,

Co-authors institute: - Genexplore Diagnostics and Research Centre Pvt. Ltd., Department of Biosciences, Veer Narmad South Gujarat University, Genexplore Diagnostics and Research Centre Pvt. Ltd., Genexplore Diagnostics and Research Centre Pvt. Ltd., Genexplore Diagnostics and Research Centre Pvt. Ltd., Genexplore Diagnostics and Research Centre Pvt. Ltd., Genexplore Diagnostics and Research Centre Pvt. Ltd., Department of Zoology, Gujarat University,

Aims: - Current study presents a compelling case that highlight the necessity of cytogenetics along with microarray technology between parental and product of conception (POC) sample. This case highlights the clinical significance of integrating both techniques to provide comprehensive genetic insights into understanding one of the reasons behind pregnancy loss.In this case study, we included a couple who had experienced recurrent pregnancy loss and sought genetic evaluation to uncover the underlying causes. Initial investigations included a POC sample which was subjected to microarray analysis as karyotype could not be done from tissue.Subsequently, karyotyping of both parents revealed that one of the parent showed an inversion of chromosomes 6.Remarkably, results of the microarray analysis of the POC sample showed a deletion and duplication of small segment in chromosome 6 consistent with the parental karyotyping. This concordance may provide some crucial information to explain the recurrent pregnancy loss in the couple.

Methods: - In our study, we conducted a microarray analysis using Agilent Platform for product of conception sample. Upon detecting abnormal results, we recommended parental karyotyping using GTG banding technique to investigate the inheritance pattern of the abnormal chromosome in the aborted fetus.

Results: - This study presents a noteworthy genetic anomaly involving chromosome 6, characterized by a 40.580 Mbp gain encompassing the region from 6p25.3 to 6p21.1, coupled with a 17.718 Mbp loss spanning the region from 6q25.2 to 6q27 suggesting an inversion of chromosome 6, based on parental karyotype. Furthermore, microarray and karyotype analysis depicted the chromosomal arrangement as 46,XX,inv(6)(p21.2q25), affirming the structural inversion's presence in the studied individual.

Conclusions: - This case study provides a clear illustration of a chromosome 6 inversion in POC with breakpoints at 46,XX,inv(6)(p21.2,q25). The combined use of microarray analysis and parental Karyotyping confirmed both its hereditary nature and the specific genetic elements impacted. This approach not only enhances our understanding of the structural variant, but also underscores its potential clinical relevance. By integrating both of this techniques, we gain valuable insights into the complexity of chromosomal rearrangements, emphasizing their significance in genetic diagnosis and counselling.

Keywords: - This study presents a noteworthy genetic anomaly involving chromosome 6, characterized by a 40.580 Mbp gain encompassing the region from 6p25.3 to 6p21.1, coupled with a 17.718 Mbp loss spanning the region from 6q25.2 to 6q27 suggesting an inversion of chromosome 6, based on parental karyotype. Furthermore, microarray and karyotype analysis depicted the chromosomal arrangement as 46,XX,inv(6)(p21.2q25), affirming the structural inversion's presence in the studied individual.