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Abstract Topic: - Molecular and cytogenetic diagnostics

Abstract Title: - The importance of conventional cytogenetics in baseline testing- a case study of complex chromosomal rearrangement

Presenting author name: - Jiju James

Presenting author institute: - Lilac Insights Pvt. Ltd.

Co-authors name: - Premkumar Torane, Ajinkya Jadhav, Vidya Bhairi, Dr. Yamini Jadhav, , , , ,

Co-authors institute: - Lilac Insights Pvt. Ltd., Lilac Insights Pvt. Ltd., Lilac Insights Pvt. Ltd., Lilac Insights Pvt. Ltd., , , , ,

Aims: - The aim of the present study is to provide an accurate diagnosis in the recurrent spontaneous abortion cases.

Methods: - We have used combination of current molecular technique, including chromosome karyotype study and chromosomal microarray study to characterized the complex chromosomal abnormalities in this patient with recurrent spontaneous abortion.

Results: - The non-consanguineous couple with the history of five missed abortions at ~8-12 weeks was referred to our centre for investigation of pregnancy loss. The chromosomal microarray study of the products of conception of sixth pregnancy showed gain of 29.2Mb on chromosome 7 in q21.13-q31.31 region and loss of 11Mb on chromosome 9 in p24.3-p23 region, loss of 8.4Mb on chromosome 21 in q11.2-q21.1 region and loss of 1.9Mb on chromosome 1 in q31.3 region.

The chromosome karyotype study of male partner showed normal 46,XY karyotype whereas the female partner showed abnormal complex chromosomal rearrangement involving five chromosomes 46,XX,t(1q;2p;7q;9p;21q).

Here we proved that combination of chromosomal microarray study is an efficient and effective workflow to identify the complex structural abnormality and to locate the rearrangement of DNA fragments.

Conclusions: - We presented a rare complex chromosomal rearrangement involving five chromosomes. The results from multiple molecular and cytogenetic techniques can provide the most comprehensive genetic analysis. This study provided detailed information for the subsequent reproductive decision making and genetic counselling for patients with recurrent spontaneous abortion.

Keywords: - The non-consanguineous couple with the history of five missed abortions at ~8-12 weeks was referred to our centre for investigation of pregnancy loss. The chromosomal microarray study of the products of conception of sixth pregnancy showed gain of 29.2Mb on chromosome 7 in q21.13-q31.31 region and loss of 11Mb on chromosome 9 in p24.3-p23 region, loss of 8.4Mb on chromosome 21 in q11.2-q21.1 region and loss of 1.9Mb on chromosome 1 in q31.3 region.

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