Abstract ID:- 210

Abstract Topic:- Molecular and cytogenetic diagnostics

Abstract Title:- Central role of conventional Karyotyping amid advance genetic technology.

Presenting author name :- Vidhi Sambhvani

Presenting author institute:- Diagnostics & Research Center Pvt. Ltd.

Co-authors name:- Jinal Parmar, Arpitsen Parmar, Alpesh Patel, Devendrasinh Jhala, Shiva Shankaran Chettiar

Co-authors institute:-Diagnostics & Research Center Pvt. Ltd., Department of Zoology, Gujarat University, Ahmedabad.

Aims:-Explore the captivating realm of cytogenetics, delving into the mesmerizing charms of genetic diversity and uniqueness. Through in-depth analysis, focus on unveiling unique karyotypes that elude identification by advanced molecular cytogenetic techniques.

Methods:- This retrospective study involved 10 distinct cases, all sourced from our database. Phytohemagglutinin-M (PHA-M) stimulated peripheral blood lymphocytes were used to obtain well spread metaphase plates for karyotyping using GTG banding technique and NOR staining.

Results:- This study identified three cases characterized by intricate balanced translocations, leading to complex chromosome arrangements: [46,XY,t(4;9;20)(q26;q31;q31.1),22ps+; 46,XX,t(2;20;6;10)(q21;p12;q13;q11.2)and 46,XX,t(1;2)(q31;q33)]. Additionally, we observed five intriguing cases, including two instances of marker chromosomes among them one was marker chromosomes with translocations [46,XY,+mar and 46,XY,t(15;21)(q10;q10),+mar]; one case of insertion [46,XX,ins(11;22)(q24;q13)] and one case of inversion [46,XY,inv(8)(p12;q21.3)].most unique case of 46,XX, inv(21)(p11.2;q21),9qh+ and one case of inversion 21 which is later confirmed by NOR staining We also encountered a case of chromosomal ring formation of the chromosome 13 [46,XY,r(13)], and a rare scenario involving loss of chromosome 22 and a derivative chromosome 14 resulting from a translocation between chromosomes 14 and 22 [45,XX,-22(der14) t(14;22)(q32.2;q11.2)]. These findings highlight the complexity and diversity of chromosomal abnormalities in the studied subjects.

Conclusions:- This data strongly suggests that these complex genetic variations are often missed by advanced genetic methods but are consistently found using traditional cytogenetics. This emphasizes the importance of the conventional approach in uncovering these hidden genetic intricacies, especially for clinical purposes and point out the requirement of more advanced molecular cytogenetic technique development which can help in the identification of these kind of chromosomal abnormalities using isolated DNA from the Patient.

Keywords:- karyotyping, Banding, translocation, inversion, insertion