عنوان مقاله:

Estimated of Chromosome Structural abnormalities inpatients passes on Genetic laboratory of Qazvin University of medical sciences

محل انتشار:

سومین کنگره بین المللی و پانزدهمین کنگره ملی ژنتیک ایران (سال: 1397)

تعداد صفحات اصل مقاله: 2

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خلاصه مقاله:

Structural chromosomal abnormalities are estimated to occur in around 0.5% of newborn infants. Chromosomes are the structures that hold our genes. If a chromosome or piece of a chromosome is missing or duplicated, there are missing or extra genes respectively. Many children with a chromosomal abnormality have mental and/or physical birth defects, ranging from mild to severe. In addition, some chromosomal abnormalities result in miscarriage or stillbirth. Chromosome disorders are of conditions, caused by constitutional numerical or structural abnormalities of chromosomes. Structural changes occur within the chromosomes themselves, not necessarily accompanied by any numerical change. There are varieties of chromosomal rearrangements that occur causing changes in the structure or components of a chromosome including: Translocations, Inversions, Ring chromosomes and Deletions.Materials and Methods: After blood sampling, culture, harvesting and preparation of metaphase spreads, we analyzed karyotype according to standard protocol in 3378 referred patients to the Genetic section of Reference Lab of Qazvin University of medical sciences during 2010-2017.Results: Our result shown 65 affected (23 translocations, 22 invesion, 4 deletion, 5 insertion, 2 ring chromosome, 14 inversion 9 (normal polymorphism) cases have chromosomal rearrangement.Discussions: Our result has shown chromosomal rearrangement were present in 1.9% of 3378 .patients. In our results, the most frequent chromosome rearrangement observed is translocation similar other studies

کلمات کلیدی:

inversions; insertions; deletions; ring chromosomes; karyotype