

Octaploidy in idiopathic thrombocytopenic purpura

R. N. Makroo, Mohit Chowdhry, Manoj Mishra, Priyanka Srivastava, Ashish Fauzdar

Department of Transplant Immunology, Molecular Biology and Transfusion Medicine Apollo Hospitals, New Delhi, India

We report a case of an elderly 68-year-old male who presented in our hospital with chief complaints of petechial rashes and ecchymosis over extremities and bleeding from the oral cavity since 3–4 days prior to hospitalization. He saw a physician before coming to our hospital and received one dose of IV methylprednisolone and oral wysolone. He had come to our hospital for further management. Bone marrow karyotyping was done and chromosomal analysis revealed two cell lines. Eighty percent of the cells analyzed revealed apparently normal male karyotype. However, 20% cells analyzed revealed a total of 184 chromosomes, suggesting octaploidy.

Key words: Idiopathic thrombocytopenic purpura, octaploidy, cytogenetics

octaploid (eight sets; 8x), decaploid (10 sets; 10x) and dodecaploid (12 sets; 12x).^[1]

We present a unique case of a chronic idiopathic thrombocytopenic purpura (ITP) patient who presented in our hospital with low platelet count. However, on bone marrow karyotyping, it was incidentally discovered that he had an octaploidy of autosomes.

Case Report

An elderly 68-year-old male, a known case of chronic ITP for more than a decade, presented in our hospital with chief complaints of petechial rashes and ecchymosis over extremities and bleeding from oral cavity since 3-4 days prior to hospitalization. He saw a physician before coming to our hospital and received one dose of IV methylprednisolone and oral wysolone. He had come to our hospital for further management. All routine investigations were done.

His initial reports revealed a very low platelet count of 1000. Vitamin B-12 was very low at 74.30. Ultrasonography of the abdomen showed splenomegaly and prostatomegaly. He was started with oral corticosteroids. The oral lesion gradually improved and there was no bleeding from any site. The platelet count was monitored regularly, and it gradually increased to around 65,000/cuml. He was also additionally started on Vitamin B-12 replacement. He was stable for 10 days, but his platelet counts dropped to around 23,000/ cuml soon after. He was then given anti-D dose, after which his platelet counts improved gradually.

Meanwhile, his bone marrow karyotyping was also done and chromosomal analysis revealed two cell lines. Eighty percent of the cells analyzed revealed apparently

Introduction

Most organisms are normally diploid, which means that they have two sets of chromosomes: one set inherited from each parent. Polyploidy refers to a numerical change in a whole set of chromosomes. True polyploidy rarely occurs in humans.^[1] Polyploidy can also be induced in cell culture by some chemicals such as colchicine.

Polyploid types are labeled according to the number of chromosome sets in the nucleus: triploid (three sets; 3x, [69,XXX]), tetraploid (four sets; 4x, [92,XXXX]), pentaploid (five sets; 5x), hexaploid (six sets; 6x),

Access this article online

Quick Response Code:



Website:

www.ijhg.com

DOI:

10.4103/0971-6866.92091

normal male karyotype. Incidentally, 20% cells analyzed revealed a total of 184 chromosomes, of which 176 autosomal chromosomes showed octaploidy with four copies each of X and Y chromosome [Figure 1].

Discussion

Idiopathic thrombocytopenic purpura

ITP is a clinical syndrome in which a decreased number of circulating platelets (thrombocytopenia) manifests as a bleeding tendency, easy bruising (purpura) or extravasation of blood from capillaries into skin and mucous membranes (petechiae).

In persons with ITP, platelets are coated with autoantibodies to platelet membrane antigens, resulting in splenic sequestration and phagocytosis by mononuclear macrophages. The resulting shortened life span of platelets in the circulation, together with incomplete compensation by increased platelet production by bone marrow megakaryocytes, results in a decreased platelet count.^[2]

Three forms of the ITP occur on the basis of duration of the disease: acute, chronic and intermittent. The chronic form of ITP is more common in young women,

with a female to male ratio of 3:1.^[2] Serum platelet autoantibodies are found in the chronic form. These antibodies are believed to be against the GPIIb/IIIa, fibrinogen receptor on the platelet membrane.^[2]

Bone arrow karyotyping

Karyotyping is the process of pairing and ordering all the chromosomes of an organism thus providing a genome-wide snapshot of an individual's chromosomes. Karyotypes are prepared using standardized staining procedures that reveal characteristic structural features for each chromosome. Clinical cytogeneticists analyze human karyotypes to detect gross genetic changes— anomalies involving several megabases or more of DNA.^[3]

The *modal number (mn)* may be expressed as a range between two chromosome numbers. Modal numbers in the *haploid (n)*, *diploid (2n)*, *triploid (3n)* or *tetraploid (4n)* range, or near but not equal to any multiple of the haploid number, and which cannot be given as a precise number, may be expressed as *near-haploid (n±)*, *hypohaploid (n-)*, *hyperhaploid (n+)*, *near-diploid (2n±)*, *hypodiploid (2n-)*, *hyperdiploid (2n+)*, *near-triploid (3n±)*, *hypotriploid (3n-)*, *hypertriploid (3n+)*, *near-tetraploid (4n±)*, *hypotetraploid (4n-)*, *hypertetraploid (4n+)*, and so

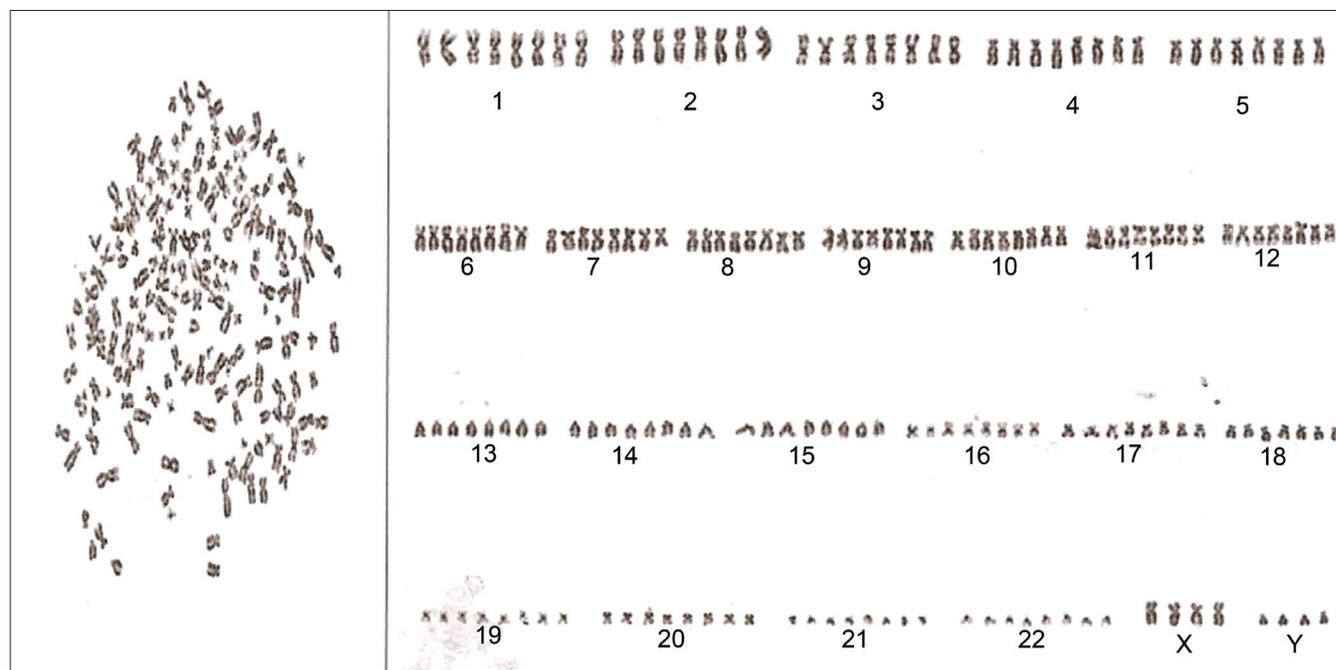


Figure 1: Metaphase and karyotype: Modal karyotype: 176,XXXX,YYYY[06]/46XY

on. Each range is determined as $n \pm n/2$, with $n/2$ defined operationally as 11 chromosomes.^[4]

Pseudodiploid, pseudotriploid, etc. are used to describe a karyotype that has the number of chromosomes equal to a multiple of the haploid number (*euploid*) but is abnormal because of the presence of acquired numerical and/or structural aberrations.^[4]

Octaploidy or near-octaploidy is a rare finding which has not been reported in a case of immune thrombocytopenia. A solitary case of near-octaploidy has been reported in a case of essential thrombocythemia by Kwong *et al*, in 1993.^[5]

In the present case, the patient was a chronic case of ITP since almost a decade. However, the patient had recurrent relapses and recurrences. On analyzing the karyotype, it was found that the patient had apparently normal karyotype in 80% of the cell lines, but 20% of the cell lines showed a near-octaploidy. A concrete evidence of an association of octaploidy with ITP is not present. It could be an incidental finding or may have been an association with the recurrent relapses and recurrences

the patient is experiencing. The report is hereby presented firstly because of its rarity in occurrence and secondly because it may have been associated with platelets disorder as both the cases reported (the present one and the one by Kwong *et al*.)^[5] were platelet disorder.

References

1. Anthony JF Griffiths, Jeffrey H Miller, David T Suzuki, Richard C Lewontin, and William M Gelbart. An introduction to genetic analysis. 7th ed. New York: W. H. Freeman; 2000.
2. Kueh YK. Adult Idiopathic Thrombocytopenic Purpura (ITP) - A prospective tracking of its natural history. Singapore Med J 1995;36:367-70.
3. O'Connor C. Karyotyping for chromosomal abnormalities. Nat Educ 2008;1:(1)
4. ISCN: An International System for Human Cytogenetic Nomenclature. In: Shaffer IG, Slovak ML, Campbell LJ, editors. Basel: S. Karger; 2009.
5. Kwong L, Chan AY, Wei D, Chan LC. Near-octaploidy in essential thrombocythemia. Cancer Genet Cytogenet 1993;65:74-5.

Cite this article as: Makroo RN, Chowdhry M, Mishra M, Srivastava P, Fauzdar A. Octaploidy in idiopathic thrombocytopenic purpura. Indian J Hum Genet 2011;17:238-40.

Source of Support: Nil, **Conflict of Interest:** None declared.

