MDS-014. Case Report: Woman With Myelodysplastic Syndrome and Complex Karyotype, Unusual Presentation in Age and Unfavorable Clinical Behavior

Juan Daniel Garza Escobar MD, Guillermo Sotomayor Duque MD,
José Luis Cedillo de la Cerda MD, Rosa Elva de León Cantú MD, Roberto Valdez Hernández MD

Instituto Mexicano del Seguro Social,
Monterrey, Nuevo León, México

Abstract

Myelodysplastic syndromes (MDS) are a group of heterogeneous pathologies that are characterized by clonal alterations in hematopoietic stem cells (HSC). MDS are usually seen in patients over 60 years of age and are accompanied by decreased ability of HSC to differentiate and enter apoptosis, producing bone marrow with increased cellularity conditioned by ineffective hematopoiesis and a degree of peripheral cytopenia. Approximately 30% of *de novo* MDS are associated with a complex karyotype, defined as the existence of 3 or more chromosomal abnormalities in the same karyotype. We present the case of a 27-year-old woman referred for pancytopenia, who has a son with aplastic anemia.

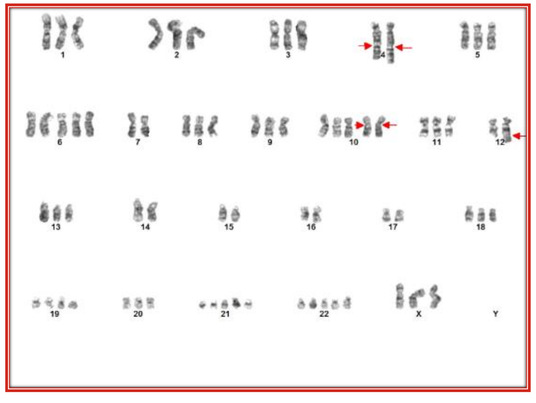
Case presentation

The patient presented a 6-month evolution picture characterized by asthenia and adynamia. She presented with pale integuments and without clinical bleeding. Initial laboratories showed hemoglobin 4.9 g/dL, hematocrit 14%, mean corpuscular volume 96.6 fL, mean cell hemoglobin 33 pg, white blood cells 2,000 cells/mL, neutrophils 900 cells/mL, lymphocytes 900 cells/mL, monocytes 100 cells/mL, platelets 36 x10^9 cells/L, lactate dehydrogenase 357 U/L. Bone marrow aspirate was performed, reporting hypercellularity for age, heterogeneous cellularity, dyspoiesis in the red series, and blasts <5%.

Flow cytometry reported 11% of total cells with $CD2^+$, $CD33^+$, $CD34^+$, $CD38^+$, $CD45^+$, $CD117^+$, and HLA^+ phenotypic expression and changes in the maturation pattern of the granulocytic series.

The Karyiotype is shown in the Fig. 1.

Fig. 1 Karyiotype



The double cell line predominated over the one that contains polyploidies, involving translocation between the long arms of chromosomes 4 and 10 and adhesion in the long arms of chromosome 12 of indeterminate origin on the normal line.

Conclusion

Myelodysplastic syndrome with excess blasts was diagnosed, and IPSS-2 high risk was concluded. During the patient's evolution, she presented a high transfusion requirement and recurrent infections, dying due to an infectious process. The patient's age is consistent with reports in the literature that MDS tend to be more aggressive at a younger age. The complex karyotype in MDS predisposes patients to clonal evolution; despite this, it is not considered a separate prognostic factor by the IPSS-R. Ideally, this group of young patients should seek early hematopoietic stem cell transplantation with curative intent.

References

1. Shahjahani, M., Hadad, E. H., Azizidoost, S., Nezhad, K. C., & Shahrabi, S. (2019). Complex karyotype in myelodysplastic syndromes: Diagnostic procedure and prognostic susceptibility. *Oncology reviews*, *13*(1), 389. https://doi.org/10.4081/oncol.2019.389
2. Veryaskina, Y. A., Titov, S. E., Kovynev, I. B., Pospelova, T. I., & Zhimulev, I. F. (2020). Prognostic Markers of Myelodysplastic Syndromes. *Medicina (Kaunas, Lithuania)*, *56*(8), 376. https://doi.org/10.3390/medicina56080376
3. Nachtkamp, K., Stark, R., Strupp, C., Kündgen, A., Giagounidis, A., Aul, C., Hildebrandt, B., Haas, R., Gattermann, N., & Germing, U. (2016). Causes of death in 2877 patients with myelodysplastic syndromes. *Annals of hematology*, *95*(6), 937–944. https://doi.org/10.1007/s00277-016-2636-z