IMAGE



Acute erythroid leukemia leading to the diagnosis of Schwachman-Diamond syndrome

Bernhard Strasser^{1*}, Sebastian Mustafa¹, Josef Tomasits² and Alexander Haushofer¹

Keywords Schwachman-Diamond syndrome, Acute erythroid leukemia, SBDS mutation



A bone marrow puncture was performed on a 38-yearold female patient with severe pancytopenia and leukoerythroblastosis (two blasts and five erythropoietic precursor cells). A cytomorphological investigation established the diagnosis of acute myeloid leukemia of the acute erythroid leukemia subtype. Myeloblasts accounted

¹ Institute of Clinical Chemistry and Laboratory Medicine, Klinikum Wels-

Grieskirchen, Grieskirchnerstraße 42, 4600 Wels, Austria

University Hospital, Linz, Austria

for 6% and erythropoietic cells accounted for 83% with a strong dominance of proerythroblasts, whereas mature hematopoietic cells were mostly absent. The proerythroblasts showed finely dispersed nuclei and minimal agranular cytoplasm. In addition, double nuclearity of erythropoietic precursor cells was frequently observed (indicated by the arrow). Immuncytological investigation provided a CD117+/71++/MPO- profile. The neoplastic cells repeatedly showed vacuolization, which is typically associated with TP53 mutation. In this patient, two somatic mutations, TP53 c.711G>A and c.706T>A, and a complex karyotype supported the diagnosis of acute erythroid leukemia.

The young age of the patient was atypical for a diagnosis of acute erythroid leukemia. Additional comorbidities included severe exocrine pancreas insufficiency. The patient exhibited growth restriction and microcephaly. A syndrome of germline predisposition to hematological neoplasms was suspected, primarily Schwachman-Diamond syndrome. Targeted sequencing of the SBDS gene revealed two heterozygote mutations, c.183_184delinsCT and c.258+2T>C, which confirmed the diagnosis of Schwachman-Diamond syndrome.

Authors' contributions

All authors contributed to the conception and design of the study. BS wrote the manuscript. AH and JT contributed to revising the manuscript. All authors read and approved the final manuscript and agree to be accountable for all aspects of the work.



© The Author(s) 2024. **Open Access** This article is licensed under a Creative Commons Attribution 4.0 International License, which permits use, sharing, adaptation, distribution and reproduction in any medium or format, as long as you give appropriate credit to the original author(s) and the source, provide a link to the Creative Commons licence, and indicate if changes were made. The images or other third party material in this article are included in the article's Creative Commons licence, unless indicated otherwise in a credit line to the material. If material is not included in the article's Creative Commons licence and your intended use is not permitted by statutory regulation or exceeds the permitted use, you will need to obtain permission directly from the copyright holder. To view a copy of this licence, visit http://creativecommons.org/licenses/by/4.0/.

^{*}Correspondence:

Bernhard Strasser

Bernhard.Strasser@klinikum-wegr.at

² Institute of Clinical Chemistry and Laboratory Medicine, Kepler

Funding

None to declare.

Declarations

Ethics approval and consent to participate

Ethical approval not required.

Competing interests The authors declare that they have no competing interest.

Received: 9 December 2023 Accepted: 15 January 2024 Published online: 06 March 2024