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Opinion Article

IKZF1 Mutation's Clinical Consequences on Acute Lymphoblastic Leukemia

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DESCRIPTION

The aggressive hematological cancer Acute Myeloid Leukemia (AML) is highly genetically heterogeneous. A portion of AML has been shown to have the initiation and maintenance related with common mutations or rearrangements but its pathophysiology is still not entirely understood. The studies concentrate on the *IKZF1* mutation, though other recurrent genetic changes with one relatively lower frequency are undoubtedly also involved in this process. The transcription factor family of zinc finger DNA-binding proteins, which also includes *IKZF2, IKZF3, IKZF4* and *IKZF5*, includes IKZF1, which encodes IKAROS (PEGASUS). One protein with a length of 519 amino acids is encoded by the *IKZF1* gene, which is located at 7p12.2 on the short arm of chromosome 7. *IKZF1* has two zinc fingers at the C-terminus that are necessary for building homo- and hetero-dimerization amongst distinct *IKZF* proteins, in addition to four zinc fingers at the N-terminal that directly bind to DNA at the core motif A/GGAAA. A minimum of 16 isoforms have been discovered due to alternative splicing or intragenic deletion, with the exception of full-length *IKZF1* (IK1). Similar to IK1, IK2 and IK3 continue to have the ability to bind DNA, whereas IK4 to IK6 lose this ability and have a predominately negative impact on full-length *IKZF1*. Functionally, IKZF1 regulates the lymphoid lineage's ontogeny and homeostasis and is essential for nearly every stage of normal lymphoid differentiation. *IKZF1* modifications primarily involve deletion and mutation. *IKZF1* alterations frequently affected Acute Lymphoblastic Leukemia (ALL), where *IKZF1* deletion rather than *IKZF1* mutation was predominate, in line with its significant role in the lymphoid system. Additionally, *IKZF1* alterations were more prevalent in Philadelphia chromosome (Ph)-positive ALL and Ph-like ALL, and they gave these ALL subtypes a poor prognosis. *IKZF1* alterations, in contrast, have received less attention in AML and there aren't as many studies on them. Children with pediatric AML have been found to frequently have *IKZF1* deletion, which is brought on by monosomy 7 or 7p deletion. Moreover, *IKZF1* mutation and monosomy 7 are more likely to be present in EVI1-rearranged AML.

IKZF1 mutation was one of the rare AML mutations, and its frequency in this cohort of newly diagnosed AML patients was 4.15% (22/530). Interestingly, studies revealed one relatively high frequency of *IKZF1* mutation compared to reports from foreign countries, especially in this cohort. This behavior was possibly ascribed to two reasons: First, there were differences in the genetic backgrounds of AML among different racial groups.

Second, recent advances in sequencing depth have shown that a large number of *IKZF1* mutations with low VAF (5%) were also found in our sample. Similar to *IKZF1* mutation, *IKZF1* deletion, including monosomy 7 and focused deletion of 7p, impairs IKZF1 function. The prevalence of *IKZF1* deletion was reported to be 3.75% (11/293) in one published study, while the frequency of *IKZF1* mutation was not significantly greater than that of *IKZF1* deletion, as evidenced by the low frequency of -7/monosomy 7 in our cohort (3.20%, 17/530). *IKZF1* deletion was considerably more common in ALL than in AML, despite *IKZF1* mutation frequency in ALL being equivalent to that in AML. According to these findings, *IKZF1* deletion was the most common *IKZF1* change in ALL, whereas *IKZF1* mutation and *IKZF1* deletion were both equally common in AML. *IKZF1* mutation was therefore one major genetic change affecting *IKZF1* and one significant gene mutation in AML.

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