TITLE: SEPT6-ABL2 FUSION FOR USE IN THE DIAGNOSIS AND/OR TREATMENT OF CANCER

FIELD OF INTEREST

Biotechnology and Molecular Diagnosis

CLINICAL NEED

T-cell lymphoblastic neoplasms (T-ALL/LBL) constitute an aggressive haematological malignancy derived from immature T cells —thymocytes— in various differentiation stages, mainly affecting children and adolescent males. The incidence, physiopathology and treatment response are highly dependent on age. The incidence is higher in children, but the disease is better characterized in terms of predisposition factors and they present high cure rates, up to 90% according to some reports. Adult patients diagnosed in Europe between 2000 and 2007, however, present a relative survival at 5 years of only 39,4% (95% CI 37,9-40,9).

T-cell lymphoblastic neoplasms exhibit recurrent genetic alterations, being the deletion of CDKN2A/CDKN2B tumour suppressor loci and activating NOTCH1 mutations the most frequent ones in over 70% and 60% of ALL cases, respectively; cytogenetic abnormalities leading to activation of oncogenic transcription factors is another hallmark, commonly from rearrangement to T-cell receptor loci, as well as additional rearrangements resulting in the expression of chimeric fusion genes involving KMT2A (MLL), HOXA and tyrosine kinase genes such as ABL1.

There is an unmet medical need of finding reliable strategies focused on the early diagnosis and treatment of T-ALL/LBL. The present invention is directed to solve this problem and a new approach for the diagnosis and treatment of these diseases is herein provided.

DESCRIPTION OF THE INVENTION

The present invention refers to the medical field. Particularly, the present invention refers to a cell linecharacterized by comprising the fusion gene [SEPT6-ABL2] within its genetic material, or the fusion transcript [SEPT6-ABL2]. The present invention is alsofocused on the use of SEPT6-ABL2 fusion in a method for the diagnosis and/or treatment of cancer.

TECHNOLOGY KEYWORDS

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TYPE AND ROLE OF PARTNER

Looking for commercial partners interested in licensing.