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Frequency of NOTCH1 Mutation and its Correlation with Clinic-Haematological Features and Teatment Response in Chronic Lymphocytic Leukemia in Sudanese Patients

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INTRODUCTION

Chronic lymphocytic leukemia (CLL) is a malignant disease that mostly affects the elderly. The clinical course and presentation are extremely variable, ranging from asymptomatic to an active disease that can cause progressive complications. CLL has shown extraordinary variety in genomic and molecular characteristics. NOTCH1 has emerged as the most frequently altered gene in CLL at the time of diagnosis linked to poor outcomes and makes treating CLL more challenging.

AIMS

To determine the frequency of NOTCH1 C.7544- 7545 delCT among Sudanese patients with CLL and its correlation with clinic-haematological features and treatment response

METHOD

This was a cross-sectional study conducted in the haematology unit at Khartoum oncology hospital, Sudan, from March to December 2022, including 29 CLL patients diagnosed according to the International Workshop on Chronic Lymphocytic Leukemia (IWCLL) (March – December – 2022). NOTCH1 C.7544-7545 delCT mutation was screened using allele specific-polymerase chain reaction (AS-PCR).

RESULTS

The overall mean age was 59.13±12.13 (30-82 years). NOTCH1 C.7544-7545 delCT mutation was detected in 4 patients (13.8%). The frequency of NOTCH1 mutation was not significantly associated with age or sex (p. value > 0.05) for both. The most frequently clinical finding among the NOTCH1 mutant was lymphadenopathy (100%) followed by the splenomegaly (50%), and hepatomegaly (25%). Anaemia and thrombocytopenia were present in 100% and 50% of patients, respectively. The frequency of NOTCH1 mutation were not significantly associated with clinical or haematological features. There was no significant association between clinical stage and NOTCH1 mutations (p=0.861) or treatment response rate (p=0.525)

CONCLUSIONS

The current study showed that the frequency of NOTCH1 C.7544-7545 delCT mutations was 13.8% in Sudanese CLL patients. The frequency of NOTCH1 mutation was not correlate with age, gender, clinical-haematological features or treatment response. This needs to be confirmed by further studies.

Table.1 Characteristics of the CLL patients

Variables	CLL patients n=61 (%)
Age , mean ±SD (range) years	59.13±12.3 (30-90)
Male, no. (%)	33 (54.1%)
Female, no. (%)	28 (45.9%)
Hb g/dl, Mean±SD Median (rangre)	10.7±2.37 11 (4.9-14.6)
Anemia , (Hb g/dl M< 12.0 ; F <11)	48(78.7%)
leukocyte (x10^9/L) Mean±SD	62.62
median(Range)	27.7 (1.63-294.5)
Lymphocytosis , no. (%)	51 (83.6%)
Platelets (x10^3/L) Mean±SD	183.33
median(Range)	189.0(10.0-486.0)

Table.2 Clinical stage and treatment response of the CLL patients

Variables	CLL patients n=61 (%)
Rai stage	
Stage 0, no. (%)	4(6.6%)
Stage I, no. (%)	2 (3.3%)
Stage II, no. (%)	3(4.9%)
Stage III, no. (%)	27 (44.3%)
Stage IV, no. (%)	25(41.0%)
Treatment response	
Complete remission,no.(%)	4 (6.6%)
Partial remission, no. (%)	15 (24.6%)
Stable disease, no. (%)	26 (42.6%)
Progression no. (%)	16 (26.2 %)

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