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CIN : U85191MH2011PTC217513

To, **Cytecare Hospitals** **Report of:** **ADHARI SALEH SAEED AL ALI (26650)**	Sample ID : 2310028188 Patient ID : 1002351570 Received on : 06-07-2023 Reported on : 15-07-2023 **Ref By: Dr. Mohanraj**

ALL MULTIPLEX PANEL REPORT

Patient Name: Adhari Saleh Saeed Al Ali (26650)	**Age: 13 Yrs/ Gender: Female**
Physician Name: Dr. Mohanraj	**Disease Status: Partially treated**
Specimen Type: Bone Marrow Aspirate	**Provisional Diagnosis: ALL**

Specimen Description: Sample quality is optimum for the test. RNA conc.: 43.2ng/µl

NO.	CHROMOSOMAL ALTERATION	GENES INVOLVED	TYPE OF LEUKEMIA	RESULT
1	t(1;19)(q23;p13)	E2A(19p13) and PBX1(1q23)	ALL (5%), 20% of pre-B ALL	Not detected
2	t(4;11)(q21;q23)	MLL1(11q23) and AF4(4q21)	ALL 2-5%	Not detected
3	t(12;21)(p13;q22)	TEL(12p13) and AML1(21q22)	B-ALL 20% of ALL mainly Pediatric	**Detected**
4	t(9;22)(q34;q11)	BCR(22q11) and ABL(9q34)	CML (95% of cases), ALL (20%), AML (M1/M2) (3%)	Not detected

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Name	ADHARI SALEH SAEED AL ALI (26650)	Patient ID	1002351570

TEST DESCRIPTION:

Acute lymphocytic leukemia (ALL) is a malignancy of B or T lymphoblasts characterized by uncontrolled proliferation of abnormal, immature lymphocytes and their progenitors which ultimately leads to the replacement of bone marrow elements and other lymphoid organs resulting in a characteristic disease pattern.

Inside leukemic cells chromosomes spontaneously rearrange themselves, called a translocation. Chromosomal translocations constitute an important parameter to identify prognostically relevant subgroups in ALL. The t(9;22) (BCR/ABL) and t(4;11)(MLL/AF4) have been associated with poor prognosis, t(1;19) (E2A-PBX1) with intermediate and t(12;21) (TEL-AML1) with good prognosis.

- Reciprocal translocation t(9,22) occurs between chromosome 9 and 22,that is Philadelphia (Ph) chromosome.
- Translocation of chromosome 4 and 11 typically occurs in ALL cases. The gene that is involved in this translocation is the MLL-AF4 fusion gene.
- Chromosomal translocation t(1,19) occurs in 5% of childhood acute lymphoblastic leukemia (ALL), the specifically targets the E2A gene and produces an oncogenic E2A-PBX1 fusion protein.
- The most frequent translocation is chromosome 12 and 21, known as TEL-AML1.

METHODOLOGY:

- Polymerase Chain Reaction.

Total RNA is extracted from whole blood, cell lines, or bone marrow cells with the QIAamp Mini Kit. cDNA is synthesized in a reaction containing the isolated RNA. The cDNA is used as a template for multiplex PCR amplification reactions. Each targets are amplified by translocation specific primers using Polymerase Chain Reaction.

References:

1. Jiménez-Morales S., Miranda-Peralta E., Saldaña-Alvarez Y.,Perez-Vera P., Paredes-Aguilera R., Rivera-Luna R. et al. 2008 BCR-ABL, ETV6-RUNX1 and E2A-PBX1: prevalence of the most common acute lymphoblastic leukemia fusion genes in Mexican patients. Leuk. Res. 32, 1518–1522.

2. Armstrong S. A., Staunton J. E., Silverman L. B., Pieters R., den BoerM. L., Minden M. D. et al. 2002 MLL translocations specify a distinct gene expression profile that distinguishes a unique leukemia. Nat. Genet. 30, 41–47.

3. Shurtleff S. A., Buijs A., Behm F. G., Rubnitz J. E., Raimondi S.C., Hancock M. L. et al. 1995 TEL/AML1 fusion resulting froma cryptic t(12;21) is the most common genetic lesion in pediatric ALL and defines a subgroup of patients with an excellent prognosis. Leukemia 9, 1985–1989.

Dr.Nirmal A. Vaniawala
MD (Path. & Bact.)

Dr. Salil Vaniawala
M.Sc Ph.D.(Human Genetics)
Consulting Geneticist

Note: This test has been performed at our partner lab. The results are based on analysis performed at partner lab.

END OF REPORT