Canterbury Health Laboratories
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Acute Leukaemia - Diagnostic Report Summary

Patient Details:

TEST, PATIENT

NHI: ABCD1234 **Born:** 09Mar2006
Doctor/GP: Seuss, Dr I am **Sex:** Female

Result Details:

Clinical Data: New diagnosis Acute Leukaemia, probable ALL.

Investigated At: Presentation *(Aged: 0 yrs, 0 mths)*

Morphology: Bone Marrow, 09 Mar 2006, AB1234X.

Aspirate:
Blast cells = 96%.
Granulopoiesis = Very few granulocytes seen.
Lymphopoiesis = The marrow is almost completely replaced by blast cells (96%).
Comments = Acute Leukaemia - morphologically probably lymphoid.

Trephine:
Diagnosis = consistent with the established diagnosis of Acute Lymphoblastic Leukaemia.

Cytochemistry: No tests performed.

Immunophenotype: Bone Marrow, AB1234X, 09 Mar 06:

Mononuclear gating = 89% of total nucleated cells.
Flow cytometry analysis of the bone marrow aspirate demonstrated 45% of gated cells were immature B cells positive for CD19, CD22, CD10, CD34 (weak) and HLA-DR. These cells were negative for CD20, surface light chain immunoglobulin, surface IgM, and aberrantly negative for CD45. This population equated to approximately 40% of total nucleated cells.
The remaining gated cells comprised 12% mature polyclonal B cells, 30% mature T cells, 6% NK cells, 1% monocytes and 1% developing myeloid cells.

Comment
These results indicate bone marrow involvement with Precursor B Acute Lymphoblastic Leukaemia. Negative expression of CD45 is an aberrant phenotype and may be useful to monitor residual disease.
Note: there is a discrepancy between the number of immature cells assessed by this technique versus the morphology differential - the sample for immunophenotyping may be haemodilute and account for this discrepancy.

Cytogenetics: continues on next page.

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Cytogenetics: Bone marrow aspirate, 09 Mar 2006, AB1234X.

Chromosome analysis of 16 GTL-banded metaphase cells revealed the following complex abnormal high hyperdiploid composite karyotype in 14 cells:

62-65,XY,+X,+3,+?5,+6,+8,+9,+10,+11,+?14,+16,+17,+18,+21,+4-6mar

The remaining two metaphase cells showed a 46,XY karyotype.

Trisomy for chromosomes 3, ?5, 6, 8, 9, 10, 11, ?14, 16, 17, 18 and 21 was detected. An additional copy of the X chromosome and approximately four to six different marker chromosomes were also observed.

Please note that chromosome morphology was poor and additional structural rearrangements cannot be excluded.

High hyperdiploid karyotypes are typically associated with a good prognosis but this may vary in cases with additional structural abnormalities. FISH analysis with an MLL specific probe is recommended.

Number of metaphase cells analysed: 16

ISCN Karyotype (1995):
62-65,XY,+X,+3,+?5,+6,+8,+9,+10,+11,+?14,+16,+17,+18, +21,+4-6mar[cp14]/46,XY[2]

FISH Haematological:

Specimen type: Cultured bone marrow cells-1 day unsynchronised
Probe: Vysis LSI MLL Dual Colour, Break Apart Rearrangement

Result: FISH analysis showed an abnormal signal pattern consistent with three intact copies of MLL in 129/160 (81%) interphase nuclei analysed. A further 12/160 (7%) of interphase nuclei showed an abnormal signal pattern consistent with four intact copies of MLL. The remaining 19/160 (12%) of interphase nuclei analysed showed a normal signal pattern.

No disruption or deletion of MLL was observed in the 160 interphase nuclei analysed. However additional copies of MLL were observed in 88% of interphase nuclei. This result is consistent with the trisomy 11 identified by standard chromosome analysis. The clone with four MLL signals may represent trisomy 11 and a marker chromosome derived from chromosome 11.

ISCN Karyotype (1995):
nuc ish 1q23(5'MLLx3,3'MLLx3)[129]/11q23(5'MLLx4,3'MLLx4)[12]/11q23(5'MLLx2, 3'MLLx2)[19]

Molecular Biology: continues on next page.

Acute Lukaemia - Diagnostic Report Summary - Continued - Continued

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Molecular Biology: Bone marrow aspirate, 09 Mar 2006, AB1234X.

BCR/ABL RT-PCR P190 Breakpoint
PRIMERS - Specific for the P190 minor breakpoint Bcr/Abl rearrangement ie E1A2.
RESULT: NEGATIVE. No Bcr/Abl (p190) transcripts detected.

t(1;19)E2A-PBX1 RT-PCR
PCR PRIMERS: specific for the breakpoints in E2A between exons 13 and 14 and PBX1 between exons 1 and 2.
RESULT: NEGATIVE: No E2A-PBX1 fusion transcripts detected.

MLL-AF4 RT-PCR
PRIMERS: Designed in MLL exon 8 and AF4 exon 7, which allows detection of all known MLL-AF4 fusion gene transcripts.
RESULT: NEGATIVE. No MLL-AF4 fusion transcripts detected

TEL/AML1 RT-PCR
METHOD: RT-PCR was performed with primers specific for the breakpoints in TEL intron 5 and AML1 introns 1 and 2.
RESULT: NEGATIVE: No TEL/AML1 gene fusion transcripts were detected.

Final Classification: (WHO) Precursor B ALL: hyperdiploidy >50
ICD-03: 9836/3

Final Comments: The following may be useful in monitoring minimal residual disease:
Immunophenotyping: CD45 negative immature B cells.
Cytogenetics: FISH analysis for Trisomy 11.

Collated By: ..... ***Approved By:*** .....*(Haematologist)* ***Date:*** .....

NB: *This document is a summation of departmental reports already issued. For full details, please refer to the original reports.*