# Educational Material IQAP 1311

The blood film was taken from a 3-year-old boy hospitalized for investigations of epistaxis and fever. The complete blood count showed WBC 543 $x10^9$/L, RBC 2.78$x10^{12}$/L, Hb 4.0 g/dL, Hct 0.224 L/L, MCV 80.8 fL and platelet 61 $x10^9$/L. Peripheral blood smear confirms the elevated white cell count with the predominance of lymphoblasts, which are small to medium size, with minimal cytoplasm leading to a very high nuclear-cytoplasmic ratio (Figure 1). Nuclei display clumped to finely dispersed chromatin and indistinct nucleoli. Some lymphoblasts with nuclear clefts are also present (Figure 2).

T lymphoblastic leukaemia and B lymphoblastic leukaemia are morphologically indistinguishable. Multi-parametric flow cytometry showed that blasts express cytoplasmic CD3, CD1a, CD2, CD5, CD7 and Tdt, but any surface CD3, CD4, CD8, B cell markers and myeloid cell markers. The immunophenotypic profile was compatible with T lymphoblastic leukaemia. Cytogenetic study demonstrated a normal karyotype. Reverse transcription-polymerase chain reaction (RT-PCR) amplified the rearranged gene transcripts of *SIL-TAL1*.

It was reported in the literature that approximately 30% of T-ALL cases carry *TAL1* gene aberrations of which 25% of cases are evident of a sub-microscopic deletion of genetic materials of 90kb in length in the region 1p32 of the long arm of chromosome 1, encoding the *SIL* gene and the un-translated region of *TAL1* gene, thereby rendering the gene expression of *TAL1* under the control of promoter region of *SIL* gene. The genetic aberration of *SIL-TAL1* can only be detected by RT-PCR or fluorescence *in-situ* hybridization using the specific probes.

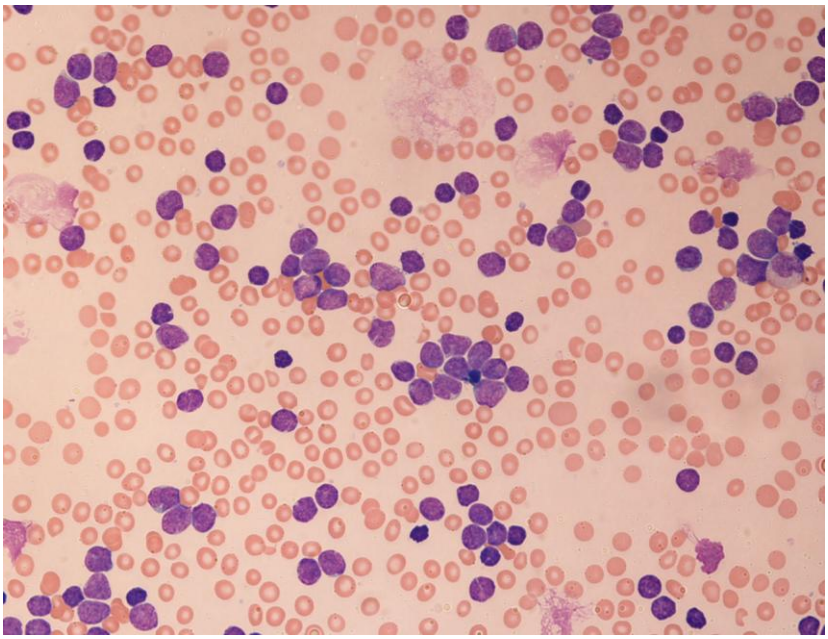

Figure 1. A predominance of lymphoblasts of small to medium size and a high nuclear-cytoplasmic ratio (400x magnification).

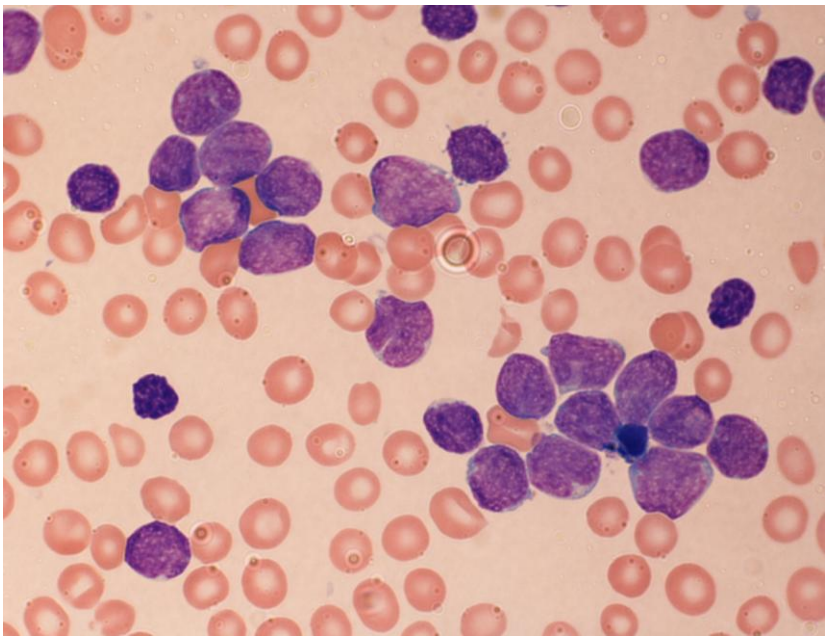

Figure 2. Lymphoblasts display clumped to finely dispersed chromatin and nuclear clefts are also seen (1,000 x magnification).