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Special Article - Hematology

Acute Myeloid Leukemia in a Patient with Shwachman-Diamond Syndrome: Complex Cytogenetics Redefined

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Shwachman-(Bodian)-Diamond Syndrome (SDS) is a rare autosomal recessive disorder (OMIM #260400): A homozygous or compound heterozygous mutation in the *SBDS* gene causes karyotypic instability, predisposing hematopoietic cells to malignant transformation. We here present a 21-year-old male patient with SDS who has had chronic leukopenia and thrombocytopenia since 4 years before presenting with pancytopenia. Bone marrow aspirate showed massive infiltration with myeloid blasts (**Figure, left panel**). Cytogenetic analysis showed highly complex chromosomal abnormalities: 97~101, XYY,-3,-5,+6,+6,+7,-8,-9,-

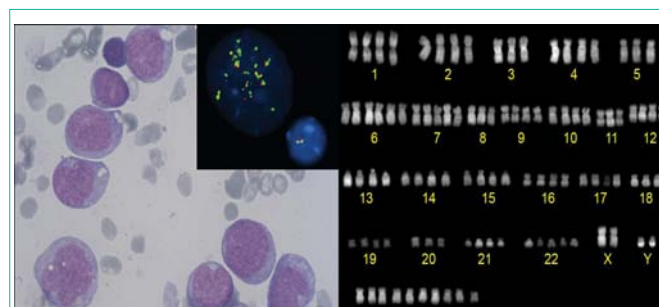


Figure: Giemsa stain, showing irregularly shaped myeloid blasts with monocytic morphology (left panel); karyogram showing multiple chromosomal abnormalities and hypertetraploidy (right panel); malignant cells had >20 signals for *KMT2A* vs 2 signals in the normal cell (inset).

11,-18,+22,+7~13mar [cp6] / 46,XY [4] (**Figure, right panel**) with FISH revealing *KMT2A* (*MLL*) amplification (11q23) (**Figure inset**). The diagnosis of poor risk acute myeloid leukemia was made. The patient was treated with high-dose chemotherapy followed by a Stem Cell Transplantation (SCT) from an unrelated donor. There are ongoing discussions whether SCT should be recommended to young adults with SDS before malignant transformation. It remains to be established whether the benefits of SCT outweigh the risk of treatment-related complications.