



## The 65th ASH Annual Meeting Abstracts

## POSTER ABSTRACTS

## 201.GRANULOCYTES, MONOCYTES, AND MACROPHAGES

**Clinical Characteristics of Shwachman-Diamond Syndrome in China - Result from Childhood Bone Marrow Failure Diseases Register of China Alliance for Blood Diseases (cBMFR-CABD)**

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## OBJECTIVE

This study aimed to identify the clinical features of 73 children diagnosed with SDS from 22 centres in China.

## METHODS

Childhood Bone Marrow Failure Diseases Register of China Alliance for Blood Diseases (cBMFR-CABD) was a bidirectional multicenter cohort study that collected data from 22 paediatric blood disease treatment centres from all over China. Patients were enrolled in the sub-centres from 1.1.2011 that were aged < 18 years at diagnosis and met the diagnostic criteria for an inherited bone marrow failure disorder. The clinical data for the 73 SDS patients analyzed in this study were collected between 1.1.2011 and 31.12.2022.

## RESULTS

Among the 73 SDS children, 38 were male and 35 were female. Short stature (height < -2SD) was found in 53.4 % (39/73) patients. Chronic diarrhea was observed in 24.7 % (18/73) patients. In children evaluated for intellectual and physical malformations, 7 patients (17.3 %, 6/52) had mental retardation and 14 (20.5 %, 15/73) patients had physical deformities.

Genetic testing was available in 63 patients. seventeen (27.0 %) patients carry bi-allelic c.258+2T>C and c.183-184TA>CT SBDS mutations. Eleven patients (17.5%) are homozygous for the c.258+2T>C mutation. Four patients (6.3%) were compound heterozygous for c.258+2T>C and exon 2 deletions. One patient carries bi-allelic mutation of DNAJC21. This patient had a history of repeated infections and pancytopenia. Bone marrow smear shows multilineage dysplasia.

Bone marrow results were available for evaluation in 54 patients including 67 % (36/54) BMF, 30 % (16/54) MDS, 4 % (2/54) AML. TP53 mutation testing was available in 15 patients with eight (53.3%) patients carry TP53 mutation. Karyotype analysis was available in 36 patients with 5 patients were complex karyotype.

Among the 56 patients available for follow-up, 17 patients underwent haematopoietic stem cell transplantation, 5 patients died (one infection, three transplant-related death and one relapse). Among the 39 non-HSCT patients, 3 patients died of AML transformation.

Three-year overall survival of all patients were 91.7% (95%CI 99.5%~83.9%), BMF patients were 100.0% (95%CI 100.0%~100.0%), MDS/AML patients were 67.7% (95%CI 94.0%~41.4%).

#### CONCLUSION

Our results provide the clinical features of the largest SDS cohort in China. A high incidence of short stature was observed. Bi-allelic c.258+2T>C with c. 183-184 TA > CT SBDS mutations were the most common mutation type. HSCT is an effective treatment modality in SDS. However, AML transformation is still a major risk factor for children with SDS.

**Disclosures** No relevant conflicts of interest to declare.

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