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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)

Yang Wan[1], *Xia Qin*[2], *Shengjiang Tan*[3], *Jianping Liu*[4], *Hailong He*[5], *Xiuli Ju*[6], *Yufeng Liu, MDPhD*[7], *Yu Jie, MD*[8], *Sun Yuan*[9], *Qun Hu*[10], *Xiaoyan Wu*[11], *Rong Liu*[12], *Jie Ma*[13], *Jing Chen, MD PhD*[14], *Alan Warren*[15], *Xiaofan Zhu, MD*[16], *Xiangfeng Tang*[17]

[1] State Key Laboratory of Experimental Hematology, National Clinical Research Center for Blood Diseases, Haihe Laboratory of Cell Ecosystem, Institute of Hematology & Blood Diseases Hospital, Chinese Academy of Medical Sciences&Peking Union Medical College, Tianjin, China
[2] National Children's Medical Center Shanghai Children's Medical Center, Shanghai, China
[3] Department of Haematology Cambridge Institute for Medical Research Wellcome - MRC Cambridge Stem Cell Institute, Cambridge, United Kingdom
[4] Inner Mongolia Maternal and Child Health Hospital, Hohhot, China
[5] Department of Hematology and Oncology, Children's Hospital of Soochow University, Suzhou, China
[6] Shandong University Qilu Hospital, Jinan, China
[7] The First Affiliated Hospital of Zhengzhou University, Zhengzhou, China
[8] Children's Hospital Affiliated to Chongqing Medical University, Chongqing, CHN
[9] Beijing Jingdu Children'S hospital Co.Ltd, Beijing, CHN
[10] Tongji Hospital, Tongji Medical College, Huazhong University of Science and Tech, Wuhan, CHN
[11] Tongji Medical College, Huazhong University of Science and Technology, Wuhan, China
[12] Capital Institute of Pediatrics, Beijing, China
[13] Hematology Center, Beijing Key Laboratory of Pediatric Hematology Oncology; Nati, Beijing, CHN
[14] Key Laboratory of Pediatric Hematology & Oncology of the Ministry of Health of China, Department of Hematology & Oncology, Shanghai Children's Medical Center, School of Medicine, Shanghai Jiao Tong University, Shanghai, China
[15] University of Cambridge, Cambridge, United Kingdom
[16] State Key Laboratory of Experimental Hematology, National Clinical Research Center for Blood Diseases, Haihe Laboratory of Cell Ecosystem, Institute of Hematology & Blood Diseases Hospital, Chinese Academy of Medical Sciences & Peking Union Medical Colleg, Tianjin, China
[17] Seventh Medical Center, PLA General Hospital, Beijing, China

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 patinets with eight (53.3%) patients carry TP53 mutation.Karyotype analysis was available in 36 patinets 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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