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Disclosures: none

Contributions: YMZ, VZ and JN conceived the study. YMZ, VZ and JN analyzed data. VZ, LC, AC, GB, EF, LF, MH, JA, GL and AN provided patient material and expertise on clinical diagnostics. MH provided information from the NOPHO registry. EF provided karyotyping data. YMZ, VZ, ACS and JN wrote the paper. ACS and JN supervised the study. All authors read and approved the final manuscript.