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Acute Leukemias

# INTERIM RESULTS OF THE QUESTIONNAIRE ON PREDISPOSITION TO ACUTE LYMPHOBLASTIC LEUKEMIA IN THE AIEOP-BFM ALL 2017 PROTOCOL

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About 8–10% of pediatric cancers are associated with pathogenic variants in cancer predisposition genes. In acute lymphoblastic leukemia (ALL), suspicion of predisposition arises from a family history of early-onset tumors, prior malignancies, comorbidities, or known genetic syndromes.

Children enrolled in the AIEOP-BFM ALL 2017 protocol completed a short questionnaire (Diagnosis Form 2) to identify clinical features suggestive of cancer predisposition.

Among 1,415 patients, 208 (14.7%) met at least one criterion for suspected predisposition. Of these, 64 (31%) had a family history of early-onset cancer (<45 years). A first-degree relative was affected in 41 cases, while 18 reported ≥2 relatives with malignancies and 14 had ≥2 second-degree relatives involved. The most frequent cancers in first-degree relatives included breast (9), thyroid (6), hematologic malignancies (7; 5 ALL, 2 AML), melanoma (5), bladder (2), and brain tumors (2), alongside single cases of other solid tumors.

Fifty-two patients (25% of the 208) had a known genetic condition at enrollment. Thirty-seven carried a cancer predisposition syndrome: 30 with Down syndrome, 3 with neurofibromatosis type 1, 2 with Noonan syndrome, 1 with Shwachman-Diamond syndrome, and 1 with a germline RB1 pathogenic variant. Additional comorbidities included congenital malformations (28; 13%) and neurodevelopmental disorders (20; 9.6%).

Genetic counseling was recommended for 153 children (11% of all enrolled). Continued collection of consultation outcomes is needed to clarify predisposition conditions, support centralized counseling and genetic testing, and ensure consistent diagnostic approaches and follow-up for affected patients.