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AGE AT DIAGNOSIS OF HEMOPHILIA: RESULTS FROM THE FRENCH HEMOPHILIA COHORT (SNH)

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Few recent and large studies provided epidemiological data about the initial presentation of hemophilia. The severity of the disease and the knowledge of previous cases in the family may influence the conditions of the diagnosis. The prospective multicenter cohort study “Suivi National des Hémophiles” (SNH) provides the opportunity to investigate the question of the age at diagnosis in French hemophiliacs.

From October 1994, 39 hemophilia centers have taken part in the ongoing SNH study, based on the voluntary participation of physicians and patients. Hemophilia patients, previously treated or not treated with clotting factors are enrolled after giving informed consent. All the data are collected on standardized forms and are recorded in a computerized database. Among 1232 patients enrolled in the SNH cohort up to December 1999, the age at diagnosis was analyzed in 537 hemophiliacs (462 with hemophilia A, 75 with hemophilia B), born between January 1980 and December 1994.

The median age at diagnosis was 7.7 months in the whole population regardless of the severity of hemophilia and of the family history. Although the frequency of diagnosis before 28 days of life was greater for hemophiliacs with family history (36 % vs 16 %,  $p < 10^{-6}$ ), early diagnosis was not the rule since even in such cases, only 45% of severe hemophilia and 27% of moderate and mild diseases were diagnosed during the neonatal period. No difference was observed between hemophilia A and hemophilia B.

Age at diagnosis (months)	Proportion up to 28 days					
	Severe (<1 IU/dl)	Mild/Moderate	Total	Severe	Mild/Mod	Total
Family history	median (n)	median (n)	median (n)			
Yes	1.8 (165)	10.6 (176)	5.3 (341)	45	27	36
No	7.8 (131)	16.6 (65)	9.3 (196)	21	8	16
Total	6.0 (296)	12.4 (241)	7.7 (537)	34	22	29

Data from the French hemophilia cohort (SNH) showed that the diagnosis of hemophilia has been delayed for patients born in 80's and early 90's, even in cases with a family history of severe disease. Initial presentation of hemophiliacs should be investigated in France, in sporadic forms as well as in non sporadic forms in the aim of optimizing the diagnosis. More extensive genetic information and carrier identification might be necessary to provide earlier diagnosis in cases with family history.