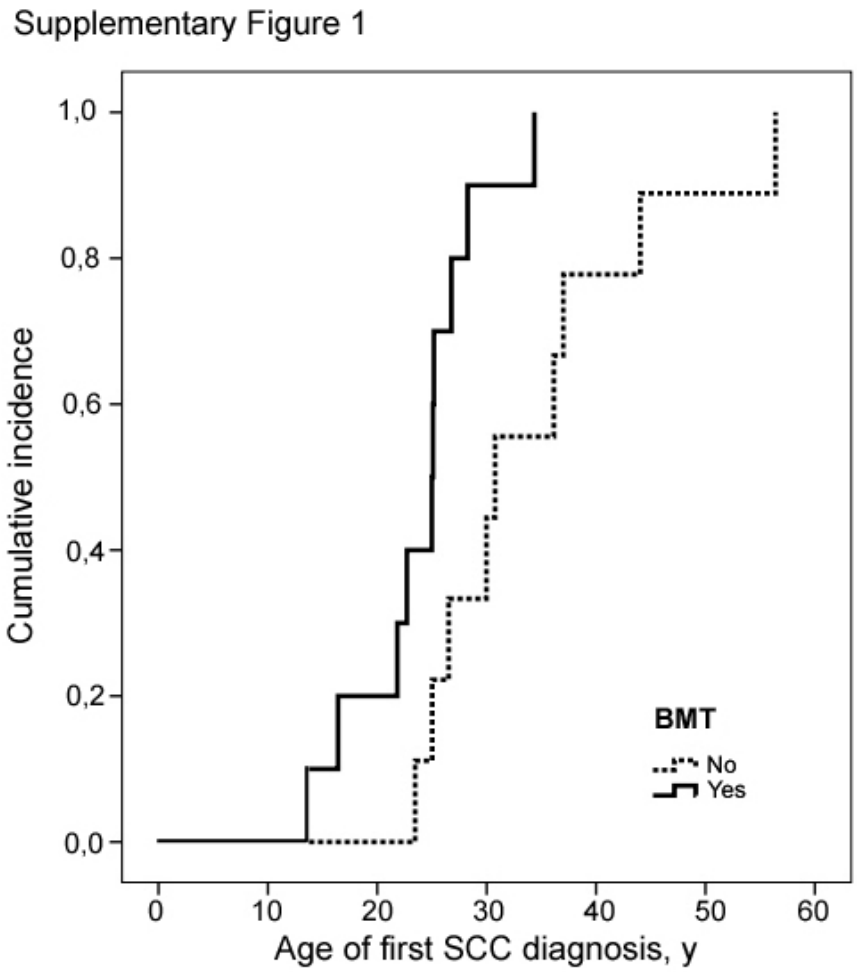
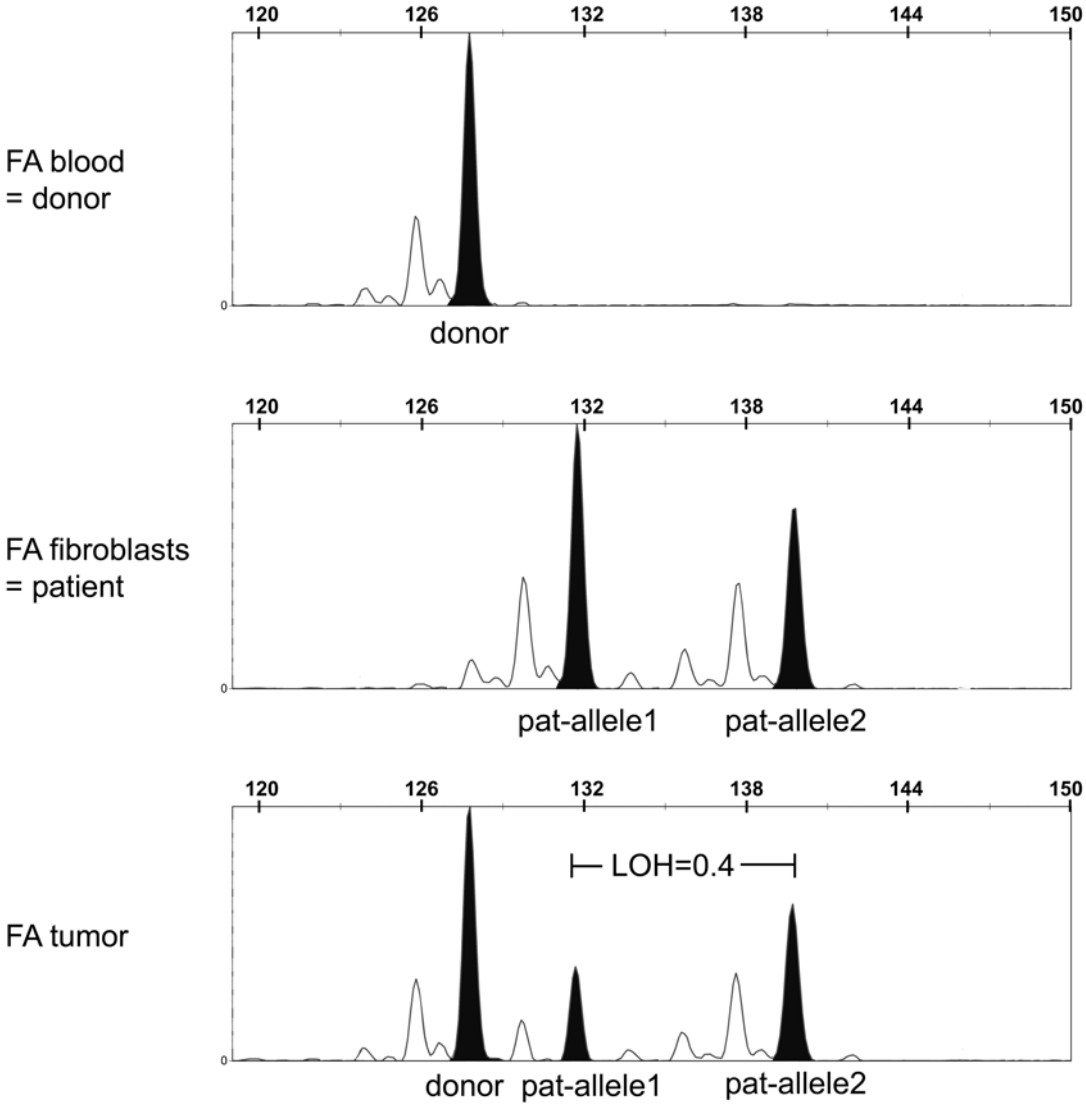
Supplementary figures



Supplementary Figure 1. **Cumulative incidence of first squamous cell carcinoma (SCC) among 19 Fanconi anemia patients with and without bone marrow transplantation.**

Incidence of SCC according to age at diagnosis of first SCC at all anatomical regions is indicated. Fanconi anemia patients who had received a bone marrow transplant were approximately 6 years younger at SCC diagnosis than those who had not received a bone marrow transplant (median age at SCC diagnosis, bone marrow transplant vs no bone marrow transplant: 24.9 years vs 30.7 years, log rank *P* = .009).

Supplementary Figure 2



Supplementary Figure 2. Analysis of cell origin (patient or donor) of Fanconi anemia (FA) squamous cell carcinomas (SCCs) in bone marrow transplanted (BMT) patients. Tumor tissue from BMT FA patients frequently showed multiple microsatellite alleles on the electropherograms: one or two arising from patient DNA, and one or two arising from donor DNA due to the presence of tumor-infiltrating donor lymphocytes. To allow analysis of the cell source of the tumor (patient or donor) even when the tumors showed mixed allelic loss patterns, we selected microsatellite markers that differed in length between the patient and the donor. The

separate alleles were usually determined in cultured fibroblasts from the FA patient (to determine patient-derived alleles) and blood of the transplanted FA patient (to determine donor-derived alleles). When the microsatellite markers of the patient and the donor were non-overlapping, they could be assigned to either the patient or the donor for assessment of allelic loss. In this example, a clear allelic loss was detected in the microsatellite alleles of the FA fibroblasts of the patient (pat-alleles1 and 2), strongly suggesting that the tumor originated from the cells of the patient (loss of heterozygosity [LOH], expressed as the ratio of the microsatellite alleles in the tumor DNA compared with those in the genetically normal DNA isolated from the FA fibroblasts, was 0.4). A change in the ratios by more than 50% (a ratio that is <0.5 or >2 relative to genetically normal DNA) is generally considered loss of heterozygosity—also called allelic loss or allelic imbalance—and typically reflects the loss of a large part of this particular chromosome). Numbers at the top of the X-axis refer to the number of base pairs indicating the allele lengths of the microsatellite marker. We had suitable material for analysis of the tumor origin in nine BMT patients. In two BMT patients (Fa12 and Fa20), we could not distinguish between the patient and the donor microsatellites, either because of a lack of pure donor and patient material or because none of the markers analyzed gave different allele lengths between donor and patient, and in these two cases we could not conclude whether the tumor was derived from patient or donor cells. In the other seven analyzed BMT patients, we could discriminate the patient-derived microsatellite markers and assessed allelic loss, thus confirming that the tumor arose in all cases from patient cells.