

Supplementary Materials

The French Cohort of DNA Repair-Deficient Xeroderma Pigmentosum Patients: Risk of Hematological Malignancies

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Table S1: Clinical and genetic description of the 181 patients of the French cohort

| Patient's code | Countries of family's origin | Sex | Ages in 2023 (years) | Ages at last seen (years) | Ages at death (years) | XP group | XP gene mutations | Clinical information |
|----------------|------------------------------|-----|----------------------|---------------------------|-----------------------|----------|-------------------|--|
| XP10VI* | Morocco | M | | | 28 | XP-C | delTG | AML-4 (27y); Death after chemotherapy at 28. |
| XP11VI* | Morocco | M | | | 25 | XP-C | delTG | Death of malignant melanoma. |
| XP14VI | French | M | | | 13 | XP-D | | XP-D/Cockayne Syndrome (CS) |
| XP22VI | North Africa | F | | 18 | | XP-C | delTG | |
| XP31VI* | Algeria | F | | 26 | | XP-C | | |
| XP32VI* | Algeria | F | | 18 | | XP-C | | |

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|----------|--------------|---|----|----|----|------|---------------------------------|---|
| XP34VI** | Gabon | M | | 31 | | XP-C | | |
| XP36VI** | Gabon | M | | 33 | | XP-C | | |
| XP51VI | Algeria | M | | 46 | | XP-V | c.1075_1244del; c.1091_1092insC | |
| XP56VI | Tunisia | F | | | 52 | XP-V | c.1075_1244del; c.1091_1092insC | |
| XP62VI | Algeria | M | | 33 | | XP-V | c.1075_1244del | |
| XP70VI | Tunisia | M | | 38 | | XP-C | | |
| XP71VI | Algeria | M | | | 14 | XP-C | delTG | Death of malignant melanoma. |
| XP75VI | Algeria | M | | 43 | | XP-V | c.1091_1092insC | |
| XP78VI | Morocco | M | | 12 | | XP-C | | |
| XP81VI* | Tunisia | M | | | 24 | XP-C | delTG | Death of malignant melanoma. |
| XP82VI* | Tunisia | M | | | 18 | XP-C | delTG | AML-6 (16y); HSCT (17y); Death of toxicity. |
| XP86VI | Algeria | M | | 53 | | XP-V | c.1561C>T | |
| XP90VI | Tunisia | F | | 24 | | XP-C | delTG | |
| XP92VI | North Africa | M | | 27 | | XP-C | | |
| XPAIHaVI | Tunisia | M | | | 25 | XP-C | delTG | Death of AML. |
| XP93VI* | Tunisia | M | 44 | | | XP-C | del TG | |
| XP251VI* | Tunisia | M | 32 | | | XP-C | delTG | |
| XP252VI* | Tunisia | M | 31 | | | XP-C | delTG | |
| XP107VI | North Africa | F | | | 17 | XP-C | | |
| XP114VI | Algeria | F | | 19 | | XP-C | | |
| XP115VI | North Africa | F | | | 25 | XP-C | | Death of malignant melanoma. |
| XP119VI | Algeria | F | | | 21 | XP-C | | |
| XP127VI | French | M | | | 57 | XP-V | c.1093dupC | |
| XP128VI | North Africa | F | | | 16 | XP-C | delTG | Leukemia (15y); Death following chemotherapy. |
| XP131VI | French | M | | | 47 | XP-D | | |
| XP134VI | North Africa | F | | 18 | | XP-C | | |
| XP146VI | French | F | | 20 | | XP-V | | |
| XP148VI | Algeria | F | | | 19 | XP-C | del TG | Poorly-differentiated follicular thyroid carcinoma (13y). |
| XP155VI | USA | M | | | 42 | XP-V | | Death of T-lymphoma following chemotherapy. |
| XP159VI | French | F | 44 | | | XP-E | c.550G>C | |
| XP162VI | Mauritania | M | | 17 | | XP-A | | XPA/CS syndrome. |
| XP163VI | Tunisia | F | | 22 | | XP-C | | |
| XP165VI | Morocco | F | | | 25 | XP-C | delTG | Kidney adenocarcinoma (23y). |
| XP166VI | North Africa | F | | | 31 | XP-C | | |

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| XP167VI | Algeria | M | | | 26 | XP-C | delTG | Trisomy 21; MDS (25y); Death of AML. |
| XP185VI | Spain | F | | | 25 | XP-C | delTG | MDS (24y); Death of AML. |
| XP188VI* | Algeria | F | 25 | | | XP-C | delTG | |
| XP189VI* | Algeria | M | 22 | | | XP-C | delTG | |
| XP204VI** | French | M | | | 29 | XP-D | c.2125C>T | Death of neurological disorders |
| XP205VI** | French | M | | | 32 | XP-D | c.2125C>T | Death of neurological disorders |
| XP208VI | Algeria | M | | | 17 | XP-C | delTG | Mediastinal lymphoma (8y). |
| XP233VI* | Tunisia | M | | | 8 | XP-C | delTG | Death of anaplastic astrocytoma. |
| XP241VI* | Tunisia | F | 30 | | | XP-C | delTG | |
| XP433VI* | Tunisia | M | 25 | | | XP-C | delTG | |
| XP235VI | Tunisia | F | | | 29 | XP-C | delTG | MDS (24); Death of AML-6. |
| XP250VI | North Africa | F | 32 | | | XP-C | | |
| XP256VI | French | F | | 48 | | XP-V | c.907C>T; c.1222_1225delACTT | |
| XP261VI* | French | F | 31 | | | XP-D | c.2125C>T; c.1925G>C | |
| XP266VI* | French | F | 27 | | | XP-D | c.2125C>T; c.1925G>C | |
| XP268VI | Greece | F | 34 | | | XP-C | c.779+1_14del12 | |
| XP269VI | Morocco | F | | | 23 | XP-C | delTG | Cervical tumor (18y). |
| XP302VI | French | M | | 20 | | XP-C | | |
| XP309VI | Morocco | F | | | 10 | XP-C | delTG | B-ALL (7y); Death of MDS following chemotherapy. |
| XP314VI*** | French | M | 30 | | | XP-C | c.1243C>T; c.1704T>A | |
| XP315VI*** | French | M | 30 | | | XP-C | c.1243C>T; c.1704T>A | |
| XP330VI | French | M | 50 | | | XP-C | | |
| XP373VI | Algeria | F | | 14 | | XP-C | delTG | |
| XP401VI | French | M | 27 | | | XP-A | | |
| XP415VI | Tunisia | M | | 29 | | XP-D | c.1802G>T | |
| XP420VI* | Morocco | F | | | 25 | XP-C | delTG | Death of cutaneous lymphoma NK. |
| XP423VI* | Morocco | F | | 36 | | XP-C | delTG | |
| XP422VI | French | F | 74 | | | XP-V | c.907C>T; c.1222_1225delACTT | |
| XP424VI | North Africa | M | 43 | | | XP-V | | |
| XP435VI* | Algeria | M | | 9 | | XP-A | c.682C>T | |
| XP436VI* | Algeria | F | | 18 | | XP-A | c.682C>T | |
| XP440VI | Brazil | F | 34 | | | XP-G | c.83C>A; c.2904G>C | |
| XP442VI | North Africa | F | | 30 | | XP-D | c.1802G>T | |
| XP456VI* | North Africa | F | 47 | | | XP-A | IVS4 C.609-2 A>G | |

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| XP457VI* | North Africa | M | 40 | | XP-A | IVS4 C.609-2 A>G | |
| XP463VI | Algeria | F | | 9 | XP-C | | |
| XP475VI* | Algeria | M | 23 | | XP-C | | |
| XP476VI* | Algeria | F | 20 | | XP-C | | |
| XP480VI | Morocco | F | 29 | | XP-C | delTG | |
| XP486VI | French | F | 72 | | XP-V | c.907C>T | |
| XP491VI | Tunisia | M | 78 | | XP-D | c.1880G>T | |
| XP492VI | Algeria | M | 59 | | XP-D | c.1802G>T | |
| XP538VI | Algeria | M | | 29 | XP-C | delTG | Death of AML-2. |
| XP546VI | Tunisia | M | 37 | | XP-V | c.1091_1092insC | |
| XP551VI | French | F | 35 | | XP-C | c.2089_2090insTGG | |
| XP552VI* | French | M | | 39 | XP-D | R683W + del Ex 3 | |
| XP553VI* | French | M | | 36 | XP-D | R683W + del Ex 3 | |
| XP574VI | North Africa | M | 31 | | XP-V | | |
| XP603VI | French | F | 75 | | XP-V | c.1727_1728delCT; c.883G>A | |
| XP606VI* | French | F | 53 | | XP-V | c.1727_1728delCT; c.883G>A | |
| XP737VI* | French | F | 57 | | XP-V | c.1727_1728delCT; c.883G>A | |
| XP612VI | Algeria | F | 24 | | XP-C | delTG | |
| XP627VI | French | M | 42 | | XP-D | L461V + del 716-730 | |
| XP629VI | North Africa | M | 26 | | XP-C | delTG | Thyroid nodules |
| XP664VI* | Tunisia | M | | 10 | XP-C | delTG | Death of astrocytoma. |
| XP665VI* | Tunisia | F | 8 | | XP-C | delTG | |
| XPAdSaVI | Morocco | M | | 19 | XP-C | delTG | Cerebellar astrocytoma (14y). |
| XP673VI | Morocco | F | | 22 | XP-C | delTG | Death of T-ALL. |
| XP689VI | Morocco | F | 22 | | XP-A | IVS4 C.609-2 A>G | |
| XP694VI | North Africa | F | 21 | | XP-C | delTG | Left Ovarian juvenile granulosa-cell tumor at 19y and right ovarian Sertoli-Leydig cell tumor at 19y. |
| XP695VI | French | F | 82 | | XP-C | c.2108C>T; c.2544delA | The oldest XP-C patient in our cohort. |
| XP700VI* | Morocco | F | 26 | | XP-A | c.682C>T | |
| XP701VI* | Morocco | F | 20 | | XP-A | c.682C>T | |
| XP704VI | French | F | 44 | | XP-C | c.1243C>T; c.1399C>T | |
| XP719VI* | Morocco | F | | 17 | XP-A | IVS4 C.609-2 A>G | Death of fulminant hepatitis. |
| XP851VI* | Morocco | M | 16 | | XP-A | IVS4 C.609-2 A>G | |
| XP731VI | North Africa | M | 43 | | XP-C | delTG | |

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| XP744VI | French | M | 20 | | XP-C | c.463C>T; c.2287delC | |
| XP757VI | French | M | | 20 | XP-D | | Hodgkin Disease (20y). Death following chemotherapy. |
| XP769VI* | North Africa | M | 23 | | XP-C | delTG | |
| XP81bVI* | North Africa | M | 20 | | XP-C | delTG | |
| XP783VI | North Africa | M | 21 | | XP-A | c.682C>T | |
| XP791VI | North Africa | M | 19 | | XP-C | delTG | A germline mutation on <i>MEN1</i> gene. |
| XP798VI* | North Africa | F | 19 | | XP-C | delTG | |
| XP855VI* | North Africa | M | 17 | | XP-C | delTG | |
| AS802VI | Algeria | M | 33 | | XP-C | delTG | Poorly-differentiated follicular thyroid carcinoma (20y). |
| XP819VI | French | M | 73 | | XP-V | c.1727_1728delCT; c.883G>A | Prostate tumor at 60y. |
| XP820VI* | North Africa | F | 19 | | XP-C | delTG | MDS/AML in January 2023 waiting for HSCT. |
| XP821VI* | North Africa | F | 21 | | XP-C | delTG | |
| XP822VI | Spain | F | 18 | | XP-D | c.1867_1868insG; c.2164C>T | Syndrome XPD/TTD. |
| XP826VI | North Africa | M | 19 | | XP-C | delTG | |
| XP843VI | North Africa | M | | 22 | XP-V | c.797T>A | |
| XP853VI | North Africa | M | | 36 | XP-V | c.278G>C | |
| XP858VI | Morocco | F | | 13 | XP-A | c.682C>T | |
| XP860VI | North Africa | F | | 29 | XP-C | c.658C>T | Death of malignant melanoma of the face. |
| XP865VI | North Africa | M | 32 | | XP-V | | |
| XP871VI | Australia | F | | 9 | XP-F | R589W + del Ex 3 31 nt | |
| XP872VI | Kosovo | F | 40 | | XP-V | c.2074A>G; c.2141A>G | |
| XP874 | Australia | M | 16 | | XP-D | c.1484T>C; c.1775G>C | Syndrome XPD/TTD. |
| XP879VI | North Africa | M | 27 | | XP-C | C.1103_1104delAA; delTG | |
| XP888VI* | North Africa | M | 36 | | XP-V | c.672delA; IVS1+1G>C | |
| XP968VI* | North Africa | F | 43 | | XP-V | c.672delA; IVS1+1G>C | |
| XP892VI | North Africa | M | 18 | | XP-C | delTG | |
| XP893VI | North Africa | M | 18 | | XP-C | delTG | |
| XP922VI | French | M | 15 | | XP-C | | |
| XP924VI* | Morocco | M | | 15 | XP-C | delTG | T-ALL (12y); MDS (13y); Death of AML-6. |
| XPEsAdVI* | Morocco | M | 18 | | XP-C | delTG | |
| XP934VI | French | F | 14 | | XP-C | c.658C>T; c.1243C>T | |
| XP961VI | French | F | 55 | | XP-V | c.437dupA | |
| XP964VI | French | F | 53 | | XP-E | | |
| XP965VI | North Africa | F | 55 | | XP-V | c.907C>T | |

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| XP966VI* | French | F | | 88 | XP-V | c.108_110del3 | |
| XP967VI* | French | F | 74 | | XP-V | c.108_110del3 | |
| XP2003VI*** | Algeria | F | 28 | | XP-C | delTG | Vaginal embryonal rhabdomyosarcoma at 16y. |
| XP2004VI*** | Algeria | F | | 23 | XP-C | delTG | Vaginal rhabdomyosarcoma at 16y and death of AML. |
| XP2006VI* | Morocco | F | 34 | | XP-C | delTG | AML-6 (29y); HSCT (29y). |
| XPToMoVI* | Morocco | M | 29 | | XP-C | delTG | |
| XPBoMoVI | North Africa | M | 32 | | XP-C | | |
| XPBoNaVI | North Africa | F | 31 | | XP-C | | |
| XPGAVI | French | F | | 54 | XP-V | c.1727_1728delCT; c.883G>A | Gastric tumor (48y). |
| XPChFaVI | North Africa | M | 28 | | XP-C | delTG | |
| XPAHVI | Tunisia | M | | 25 | XP-C | delTG | AML (24y). Curietherapy (24y). Death at 25. |
| XPChFa2VI | North Africa | M | | 24 | XP-C | | Death of leukemia. |
| XPCoJoVI | French | F | 29 | | XP-C | | |
| XPMoAbVI | North Africa | M | 35 | | XP-C | | |
| XPBeDeVI | North Africa | F | 11 | | XP-C | | |
| XPBoIlVI | North Africa | M | 17 | | XP-D | c.A254G | |
| XP2020VI | Tunisia | F | 14 | | XP-C | delTG; c.G850T | Ovarian Sertoli-Leydig cell tumor (11y). |
| XPDuMeVI | North Africa | F | 18 | | XP-C | | |
| XPRaWaVI* | North Africa | M | 10 | | XP-C | delTG | |
| XPRaYaVI* | North Africa | F | 13 | | XP-C | delTG | |
| XPAAVI | Algeria | F | 29 | | XP-C | delTG | Papillary thyroid carcinomas (17y) and pancreatic tumor (29y) under chemotherapy. |
| XPElHaVI* | North Africa | F | | 22 | XP-C | delTG | Ovarian sarcoma (18y). |
| XPElKaVI* | North Africa | F | | 28 | XP-C | delTG | Uterine adenomyosarcoma (15y). Death at 28. |
| XPMaAbVI | Morocco | M | 22 | | XP-C | delTG | AML-3 (14y). |
| XPGaViVI* | Algeria | F | | 25 | XP-C | delTG | Death of RAEB-2. |
| XPGaMVI* | Algeria | M | | 27 | XP-C | delTG | Death of RAEB-t. |
| XP1BEVI | Belgium | M | 27 | | XP-V | | |
| XPWaVI | Pakistan | M | 33 | | XP-C | c.1243C>G; c.1934delC | Angiosarcoma of the inner canthus of the left eye (22y). |
| XPAblLyVI | North Africa | F | 28 | | XP-C | | |
| XP844VI | Comoros | M | 19 | | XP-C | IVS 12-1G>C | |
| XP845VI* | Comoros | M | 27 | | XP-C | IVS 12-1G>C | |
| XPMYVI* | Comoros | F | | 30 | XP-C | IVS 12-1G>C | Death of breast tumor at 30y. |
| XP936VI | Comoros | M | 22 | | XP-C | IVS 12-1G>C | |

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|----------|---------|---|----|----|------|-------------|------------------------------|
| XPYolsVI | Comoros | M | 25 | | XP-C | IVS 12-1G>C | |
| XPMa01VI | Comoros | F | 31 | | XP-C | IVS 12-1G>C | |
| XPMa02VI | Comoros | M | 24 | | XP-C | IVS 12-1G>C | |
| XPMa14VI | Comoros | M | 22 | | XP-C | IVS 12-1G>C | |
| XPMa06VI | Comoros | F | | 32 | XP-C | IVS 12-1G>C | Death of malignant melanoma. |
| XPMa09VI | Comoros | F | 21 | | XP-C | IVS 12-1G>C | |
| XPBANaVI | Comoros | F | 21 | | XP-C | IVS 12-1G>C | Trichoblastoma. |

- (*) or (**): siblings of the closest XP patient in the list; (***): twins.
- HSCT: hematopoietic stem cell transplant; RAEB; refractory anemia with excess blasts.
- delTG means c.1643_1644 delTG; p.Val548AlafsX572 *XPC* mutation.
- CS means Cockayne Syndrome which is a DNA repair-deficient disease with neurological abnormalities and early aging but no increased risk of tumors.
- TTD means trichothiodystrophy which is a DNA repair-deficient disease with neurological abnormalities but no increased risk of tumors.