**Supplementary information**

**S7 Table. Demographic data, data for chromosomal breakage investigation and FANCD2 immunoblot for FA patients with mutations not known**

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| --- | --- | --- | --- |
| **Age** | **Gender** | **Chromosomal breakage score** | **FANCD2 Immunoblotting** |
| 12 | F | 9.48 breaks/ metaphase | S-form FANCD2 only |
| 11 | M | 4.54breaks/metaphase | S-form FANCD2 only |
| 8 | F | 13.92breaks/metaphase | S-form FANCD2 only |
| 7 | F | 7.83breaks/metaphase | S-form FANCD2 only |
| 5.5 | M | 1.89breaks/metaphase | S-form and L-form of FANCD2 |
| 15 | F | 4.69breaks/metaphase | S-form FANCD2 only |
| 14 | M | 2.23breaks/metaphase | S-form and L-form of FANCD2 |
| NA# | M | 3.44breaks/metaphase | S-form FANCD2 only |

# Age of this patient was not available.