



Fig. S1. Diagnostic flowchart for males with suspected incontinentia pigmenti. In order to achieve correct molecular genetic diagnosis, DNA from both ethylenediaminetetraacetic acid (EDTA) blood and affected tissue might be necessary. The latter should be obtained from a fresh or snap-frozen tissue sample, not paraffin-embedded tissue. Consequently, the tissue sample for molecular analysis should be obtained simultaneously with the biopsy for histological examination in order to avoid re-biopsy. It is important that the tissue sample is achieved early in the course of disease, before the affected cells bearing the mutated gene, are selectively eliminated. A 47,XXY karyotype should be diagnosed/excluded by cytogenetic analysis performed on lithium-heparin (Li-He) blood; i.e. both Li-He blood and EDTA blood should be obtained simultaneously (11).