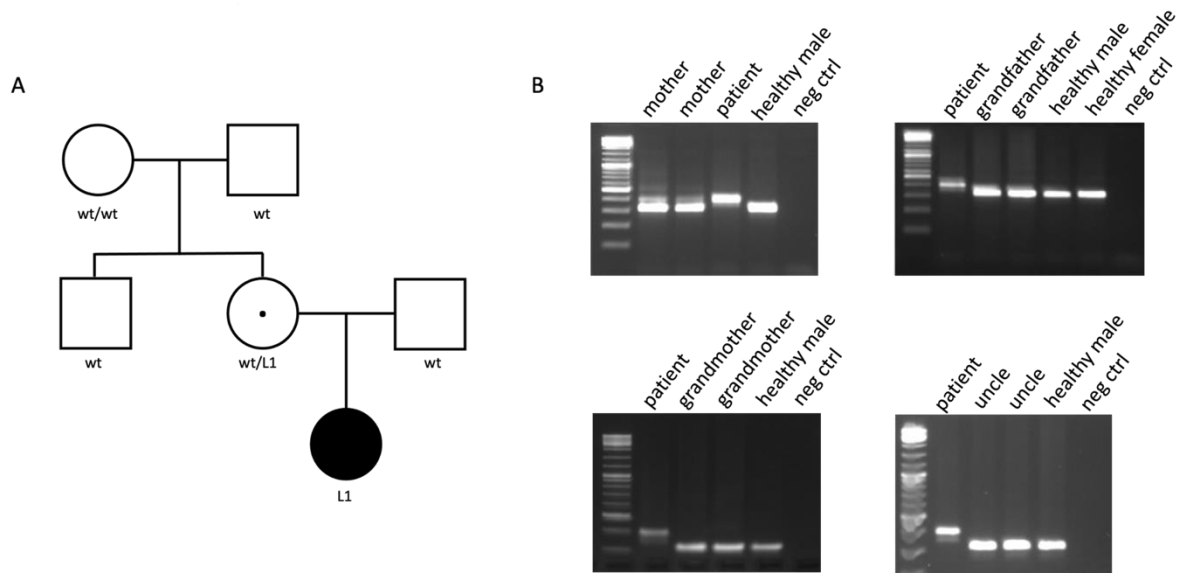
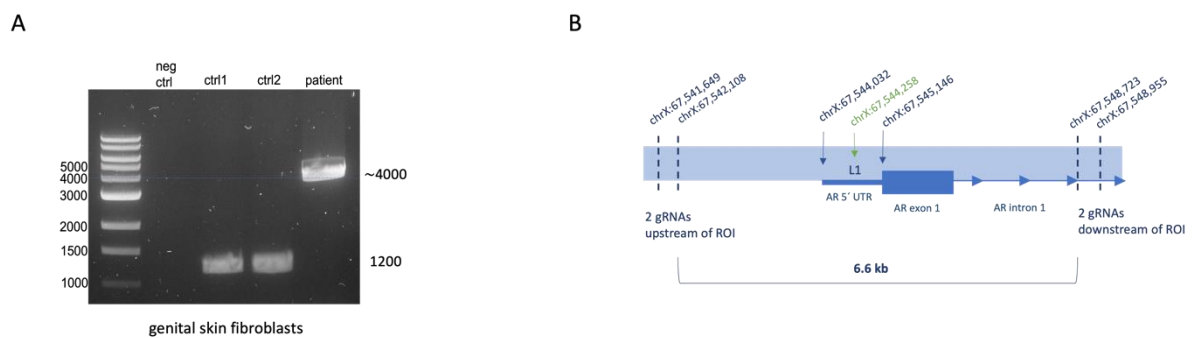


# LINE1-mediated epigenetic repression of androgen receptor transcription causes androgen insensitivity syndrome

## Supplementary Information



**Supplementary Figure 1.** Segregation of the L1 insertion in the family. (A) pedigree showing the patient (46,XY but raised as a girl), the unaffected mother (carrier) and her family. (B) PCR on blood DNA from the patient (396bp), different maternal family members and healthy controls (314bp).

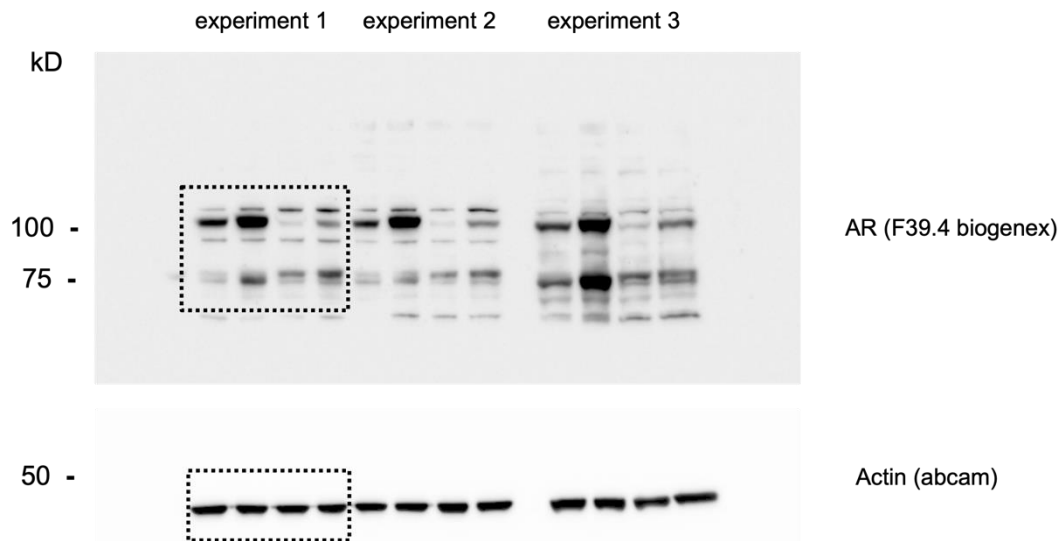


**Supplementary Figure 2.** Genetic analysis in patient-derived cells. (A) long-range PCR showing an estimated length of the L1 insertion in patient-derived cells, as compared to wild type bands in two healthy controls. (B) experimental design of Cas9-mediated target enrichment, showing target locations of gRNAs upstream and downstream of the region of interest (ROI), along with genomic coordinates in the hg38 assembly.



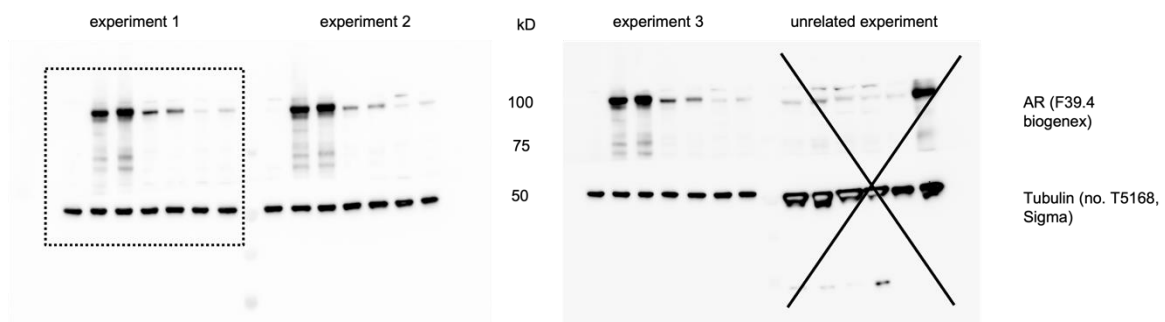
## Uncropped images

Western blot image from Figure 1:



Membrane was cut after blotting between 50 kD and 75 kD. The upper part was incubated with the anti-AR antibody F39.4 from Biogenex, the lower part was incubated with the anti-actin antibody from Abcam. Shown are three biological replicates. Dotted lines indicate the selection taken for Figure 1.

Western blot image from Figure 3:



Membrane was cut after blotting between 50 kD and 75 kD. The upper part was incubated with the anti-AR antibody F39.4 from Biogenex, the lower part was incubated with the anti-tubulin antibody from Sigma. Shown are three biological replicates. Dotted lines indicate the selection taken for Figure 3.

Agarose gel images from Supplementary Figure 1 were generated in a diagnostic laboratory according to ISO15189 guidelines.

Agarose gel from Supplementary Figure 2:

