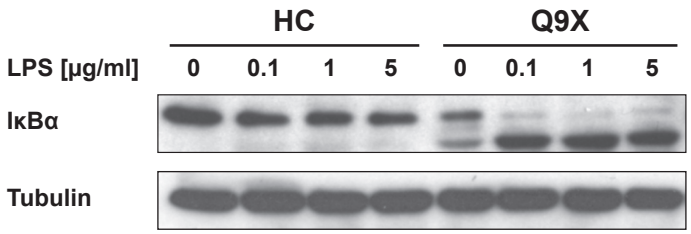
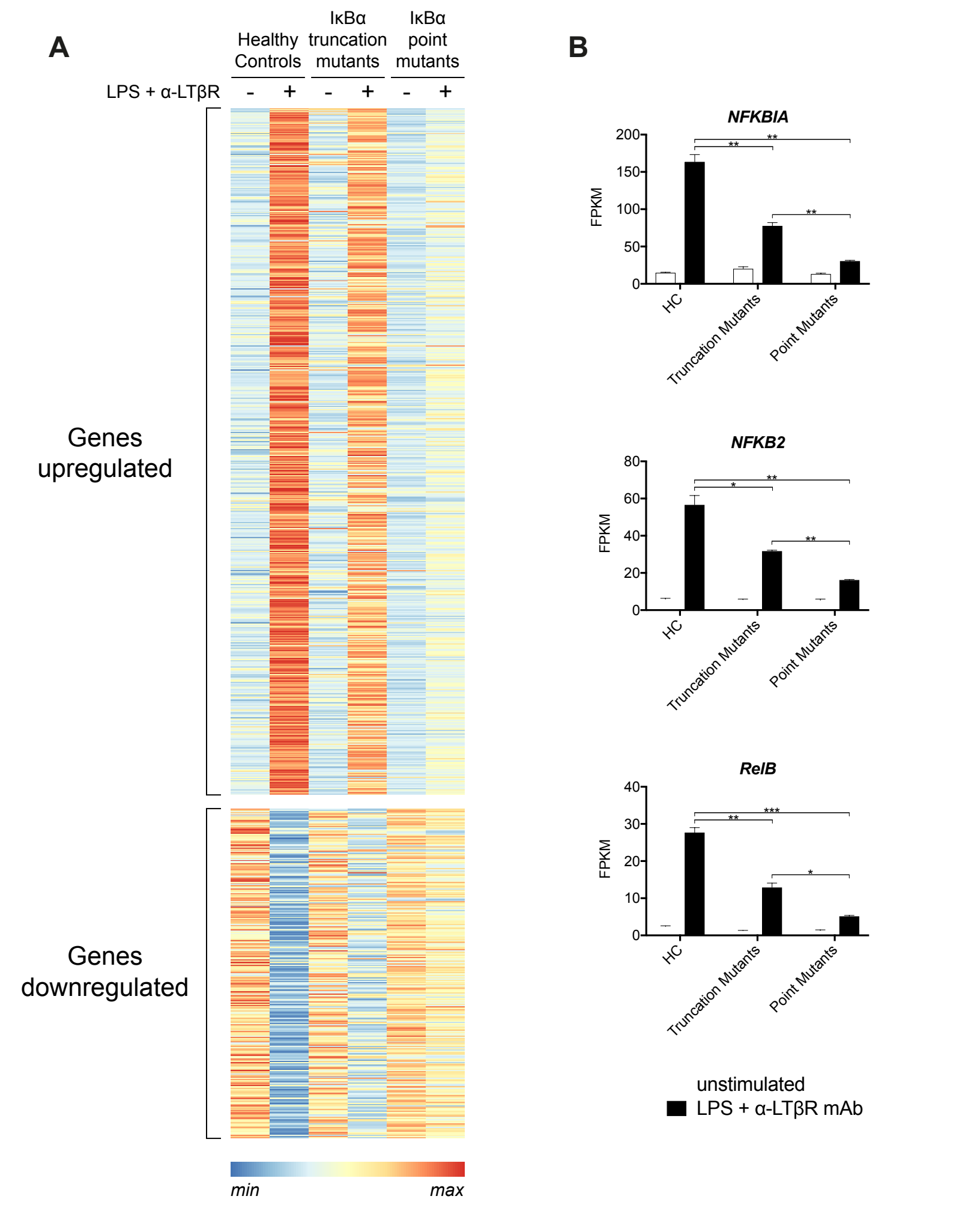


Supplemental Figure S1



Supplemental Figure S2

Supplemental Table E1. Breakdown of the disease severity score of the patients.

Patient	Infections	Ectodermal dysplasia: systems affected	Antibody responses	Lymphocyte count per µl	Memory T and/or B cells	Cumulative score
1	3	3	3	3	3	15/15
2	3	3	3	3	2	14/15
3	3	3	3	3	3	15/15
4	3	3	2	3	3	14/15
5	2	2	3	1	3	11/15
6	2	3	3	1	3	12/15
7	3	2	n.a.	1	0	6/12
8	1	2	1	0	0	4/15
9	2	1	n.a.	1	0	4/12

n.a., not available.

Genotype-phenotype correlation in autosomal dominant anhidrotic ectodermal dysplasia with immune deficiency: More severe disease and greater impairment of NF-κB activation in IκBα point mutants versus truncation mutants

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SUPPLEMENTAL FIGURE LEGENDS

Figure S1. Effect of LPS stimulation on IκBα levels in the patient with the IκBα Q9X truncating mutation. Representative IκBα immunoblot of fibroblasts from a healthy control (HC) and the patient. Two lower molecular weight IκBα bands are evident in unstimulated fibroblasts from the patient; only the band with the lowest molecular weight increased in intensity following stimulation with increasing concentrations of LPS.

Figure S2. IκBα point mutants impair more than IκBα truncation mutants the induction of various genes including those regulated by NF-κB. A. Heat map of genes the expression of which was significantly altered by LPS+anti-LTβR stimulation of normal fibroblasts. **B.** RNA-Seq analysis of the expression of *NFKBIA, NFKB2* and *RELB* mRNA in unstimulated and LPS+anti-LTβR mAb stimulated fibroblasts from healthy controls (HC; n=3) and patients with IκBα truncation (n=2) and point mutations (n=2). Results are expressed as Fragments per Kilobase of Exon per Million (FPKM). Columns and bars represent the mean±SD for each of the three groups. *, $p<0.05$; **, $p<0.01$; ***, $p<0.001$.