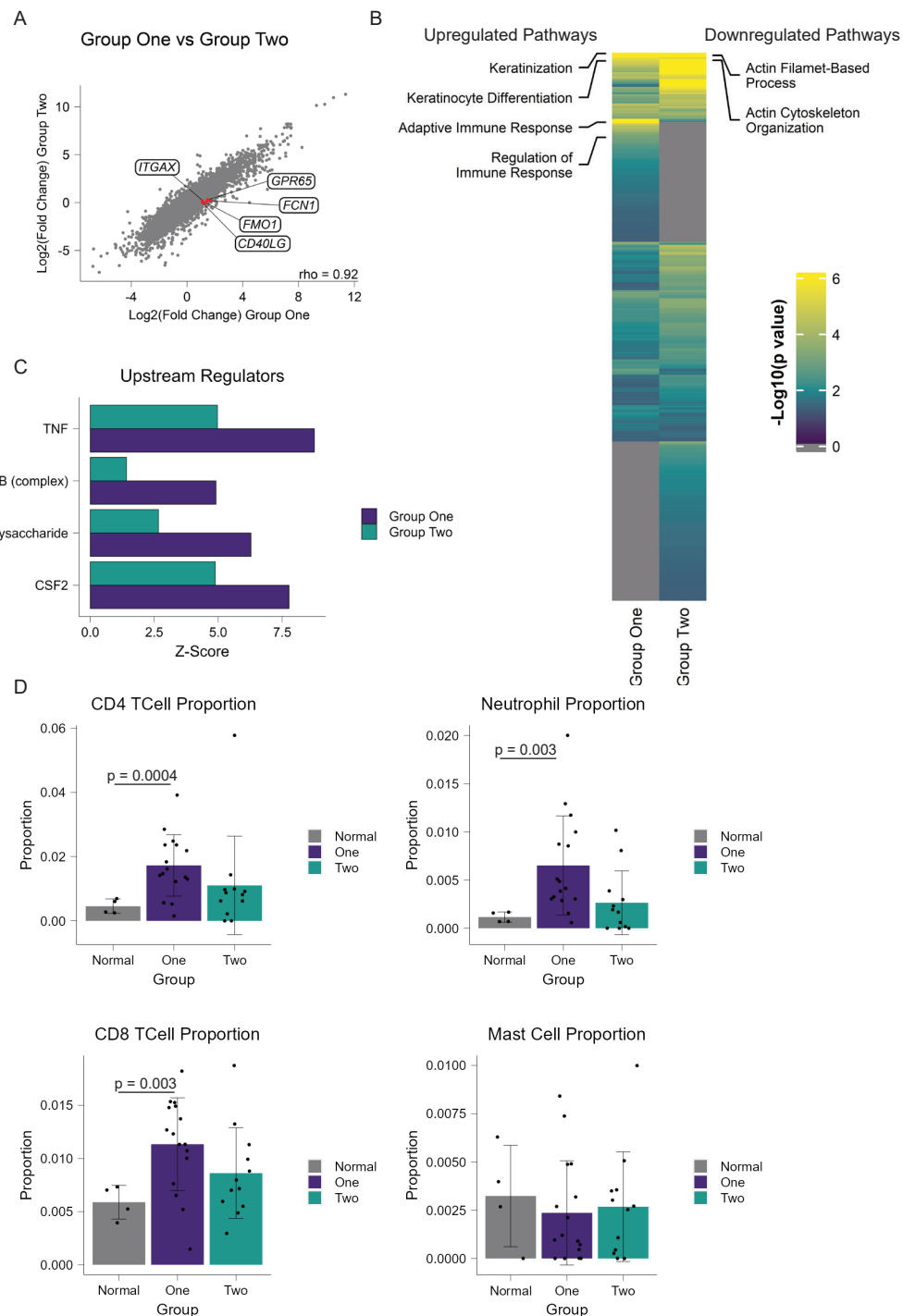


## **Supplemental Material**

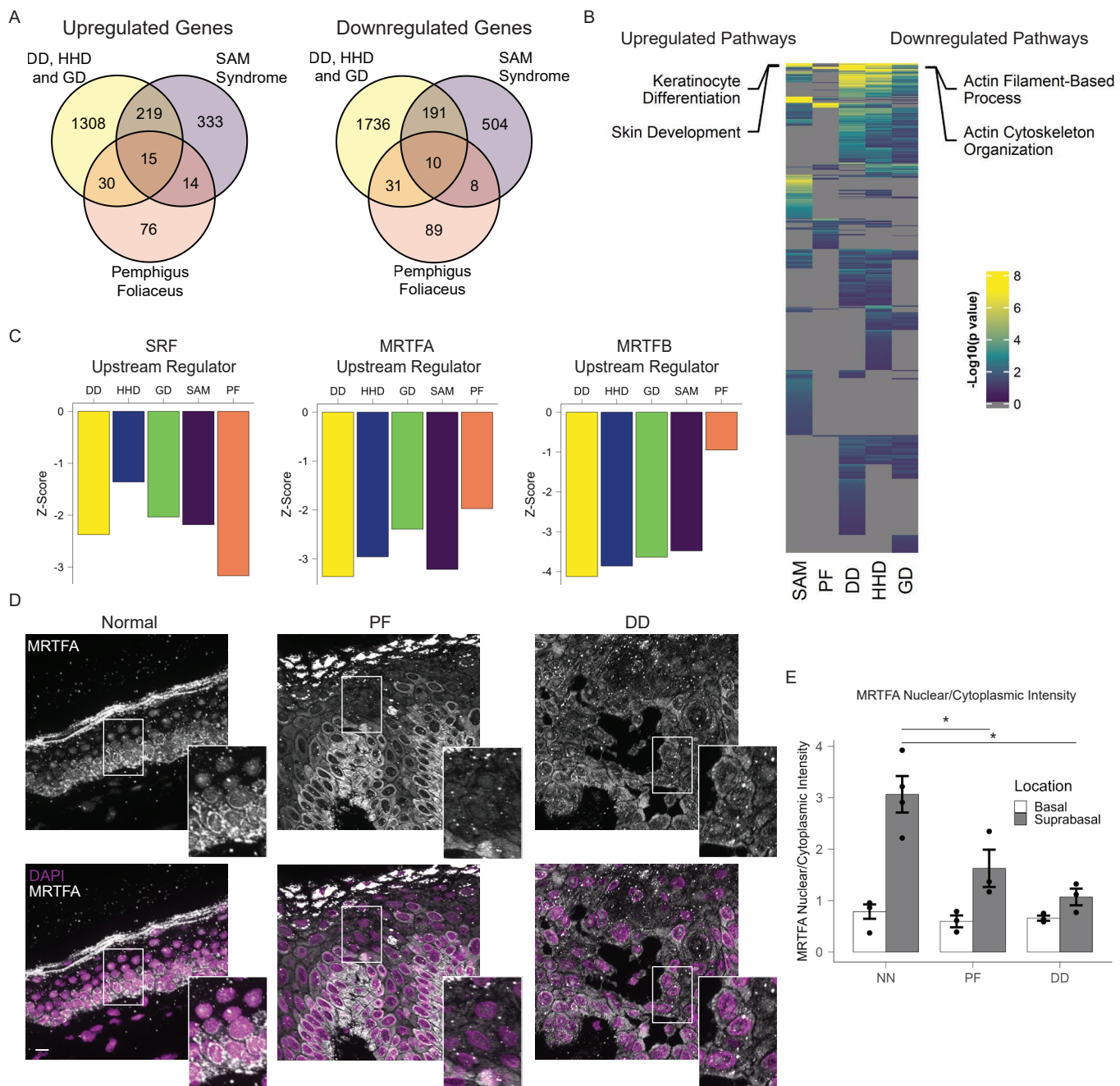
### **Transcriptional profiling of rare acantholytic disorders suggests common mechanisms of pathogenesis**

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**Supplemental Figure 1. DD, HHD and GD form two intermixed groups separated by level of inflammatory signatures.** A) Correlation analysis of gene expression from group one and group two samples. Labeled genes are the top 5 differentially ranked genes from the spearman correlation, after filtering out lowly expressed genes and for genes that are significantly changed in at least one group. B) Heatmap showing GO BP pathways ranked by  $-\log_{10}(p \text{ value})$  for group one and group two. Selected upregulated pathways are noted left of the heatmap, while downregulated pathways are noted on the right. C) Z-score from IPA upstream regulator analysis, with a focus on inflammatory mediators. D) Predicted proportion of select inflammatory cells identified using CIBER-SORTx.



**Supplemental Figure 2. Pemphigus foliaceus and SAM syndrome share changes in actin organization pathways with DD, HHD and GD.** A) Venn diagram showing overlap in upregulated genes and downregulated genes in PF, SAM syndrome, DD, HHD, and GD samples. B) Heatmap showing GO BP pathways ranked by  $-\log_{10}(\text{p value})$  for PF, SAM syndrome, DD, HHD and GD. Selected upregulated pathways are noted on the left of the heatmap, while downregulated pathways are noted on the right. C) IPA upstream regulator analysis showing z-score for SRF, MRTFA and MRTFB in PF, SAM syndrome, DD, HHD, and GD samples. D) Immunostaining for MRTFA in PF, DD, and NN skin. Quantification of MRTFA nuclear and cytoplasmic pixel intensities measured in basal and suprabasal cells. Scale bar = 20  $\mu\text{m}$ .

| Diagnostic Group | Age | Sex | Body Site                                                           |
|------------------|-----|-----|---------------------------------------------------------------------|
| Grovers          | 72  | f   | Skin of right lower back, punch                                     |
| Grovers          | 84  | m   | Skin of left abdomen, punch biopsy                                  |
| Grovers          | 76  | m   | Skin of right flank, shave                                          |
| Grovers          | 67  | f   | Skin of right upper arm, punch biopsy                               |
| Grovers          | 58  | f   | B. Skin of right mid submammary area – superior, wedge biopsy:      |
| Grovers          | 77  | m   | Skin of left chest, punch                                           |
| Grovers          | 82  | f   | Skin of mid back, punch biopsy                                      |
| Grovers          | 57  | f   | Skin of right lower back, punch                                     |
| Grovers          | 39  | f   | Skin of right arm, punch                                            |
| Grovers          | 82  | m   | B. Skin of left middle back, shave                                  |
| Hailey Hailey    | 40  | m   | B. Skin, right axilla, punch                                        |
| Hailey Hailey    | 81  | f   | Skin, right back T12, punch                                         |
| Hailey Hailey    | 45  | f   | Skin of left inframammary fold, punch                               |
| Hailey Hailey    | 47  | f   | Skin of left upper abdomen                                          |
| Hailey Hailey    | 55  | m   | Skin of central chest, wide local excision                          |
| Hailey Hailey    | 54  | f   | Skin of left antecubital fossa, punch                               |
| Hailey Hailey    | 44  | f   | Skin of left inframammary region, punch                             |
| Dariers          | 24  | m   | Skin of right upper back, shave                                     |
| Dariers          | 34  | f   | Skin of right back, punch                                           |
| Dariers          | 49  | m   | Skin of right upper back, punch                                     |
| Dariers          | 30  | f   | Skin of mid chest, punch                                            |
| Dariers          | 67  | m   | Skin of central lower back, shave                                   |
| Dariers          | 37  | m   | Skin of left shoulder, punch                                        |
| Dariers          | 45  | f   | Skin of right upper abdomen, punch biopsy                           |
| Dariers          | 61  | m   | Skin of right forearm, shave                                        |
| Dariers          | 49  | m   | Skin of left upper chest, shave                                     |
| Dariers          | 49  | m   | Skin of left malar area 5.5 cm from ala, 7 cm from tragus, excision |
| Dariers          | 20  | f   | Trunk                                                               |
| NN               | N/A | f   | Flank                                                               |
| NN               | N/A | f   | Flank                                                               |
| NN               | N/A | f   | Flank                                                               |
| NN               | N/A | f   | Flank                                                               |

**Supplemental Table 1. Detailed characteristics of individual patient biopsy samples**

| Group   | Mean Age | Female Sex |      | Male Sex |      | DD     |         | HHD    |          | GD     |         |
|---------|----------|------------|------|----------|------|--------|---------|--------|----------|--------|---------|
|         |          | Number     | %    | Number   | %    | Number | % of DD | Number | % of HHD | Number | % of GD |
| Group 1 | 53.56    | 10         | 0.63 | 6        | 0.37 | 6      | 0.55    | 3      | 0.43     | 7      | 0.70    |
| Group 2 | 55.67    | 5          | 0.42 | 7        | 0.58 | 5      | 0.45    | 4      | 0.57     | 3      | 0.30    |

**Supplemental Table 2. Characteristics of samples between group 1 and group 2.**