SUPPLEMENTARY MATERIAL

An ALMS1 variant disrupts proximal centriole organization and promotes a myofibroblast-like phenotype that is regulated by THY1

Angela C. Zeigler [1,2§], Sarah Colijn [3§], Ankur A. Gholkar [4], Song Yang [5], Xuedong Kang[1,2], Yan Zhao[2,6], Charlotte Wolf [2], Song Li [5,6], Jorge Z. Torres [4], Stanley F. Nelson [1,7], Amber N. Stratman [3], Marlin Touma *[1,2]

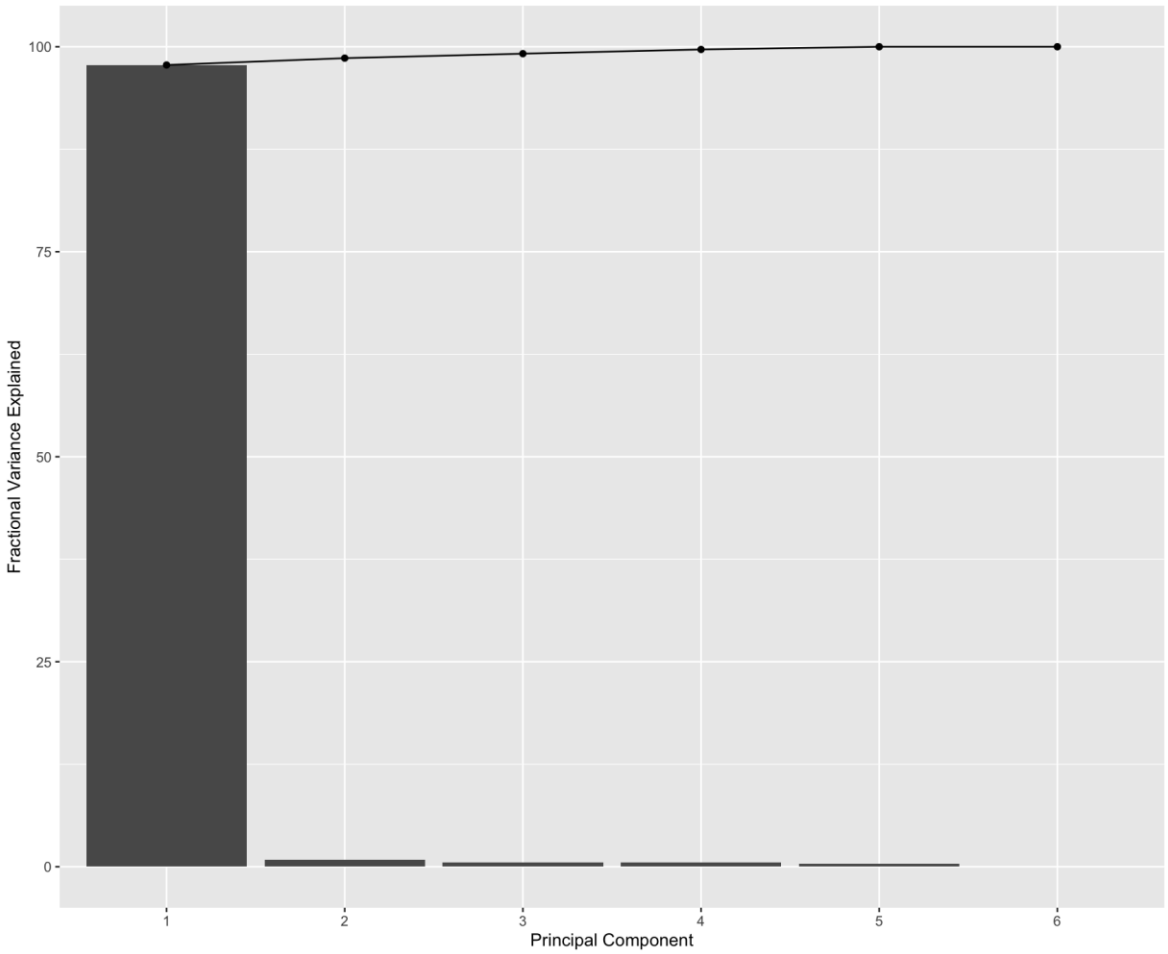
Authors' Affiliations

1. Department of Pediatrics, David Geffen School of Medicine, University of California, Los Angeles, CA.
2. Neonatal/Congenital Heart Laboratory, Cardiovascular Research Laboratory, David Geffen School of Medicine, University of California, Los Angeles, CA.
3. Department of Cell Biology and Physiology, Washington University in St. Louis School of Medicine, St. Louis, MO.
4. Department of Chemistry and Biochemistry, University of California, Los Angeles, CA
5. Department of Bioengineering, University of California, Los Angeles, CA.
6. Department of Medicine, David Geffen School of Medicine, University of California, Los Angeles, CA
7. Department of Human Genetics, David Geffen School of Medicine, University of California, Los Angeles, CA.

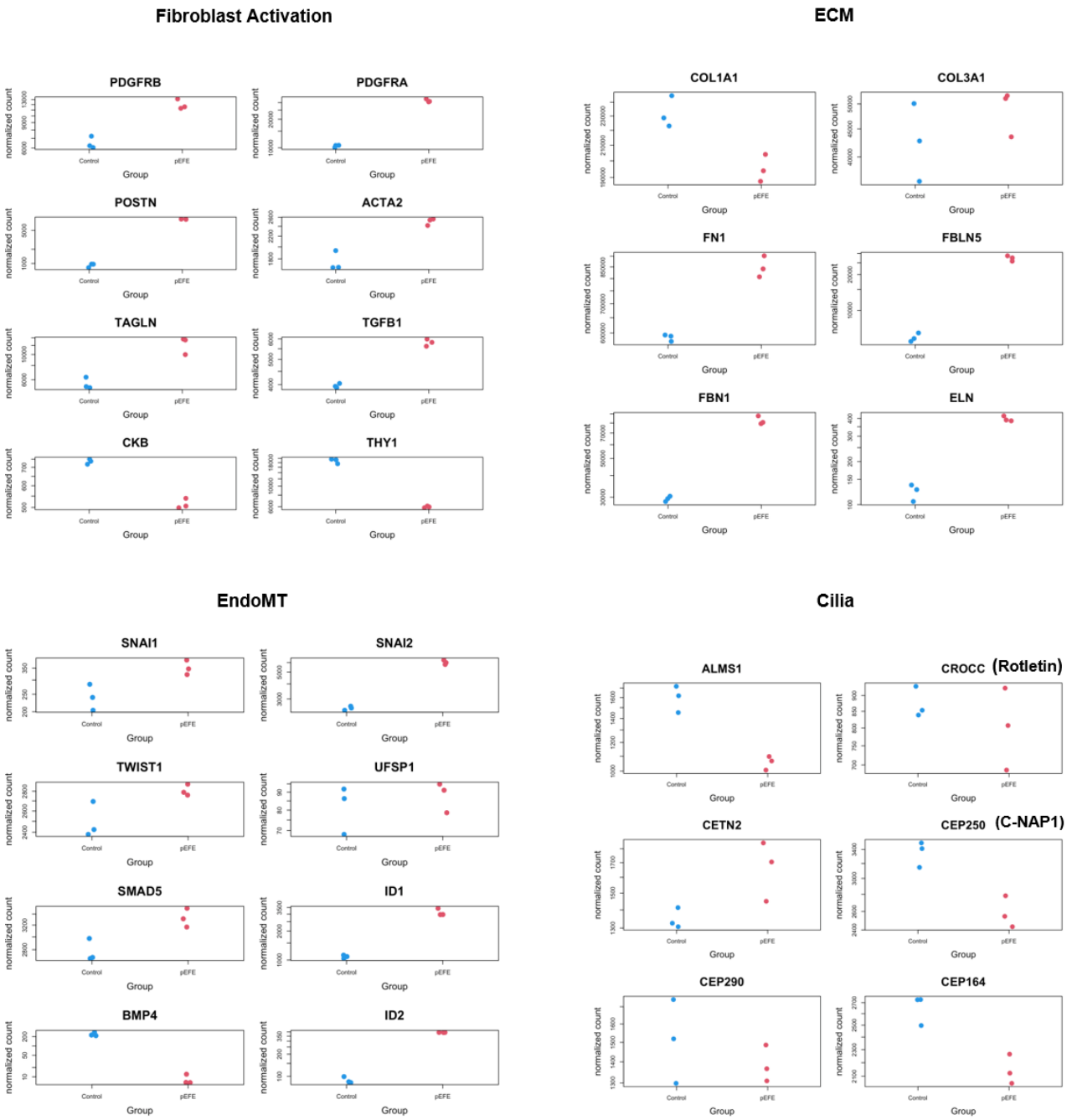
§Authors S.C. and A.C.Z. contributed equally to this work

*Correspondence:
Marlin Touma, MD, PhD
Phone: 310.206.6197
Email: mtouma@mednet.ucla.edu

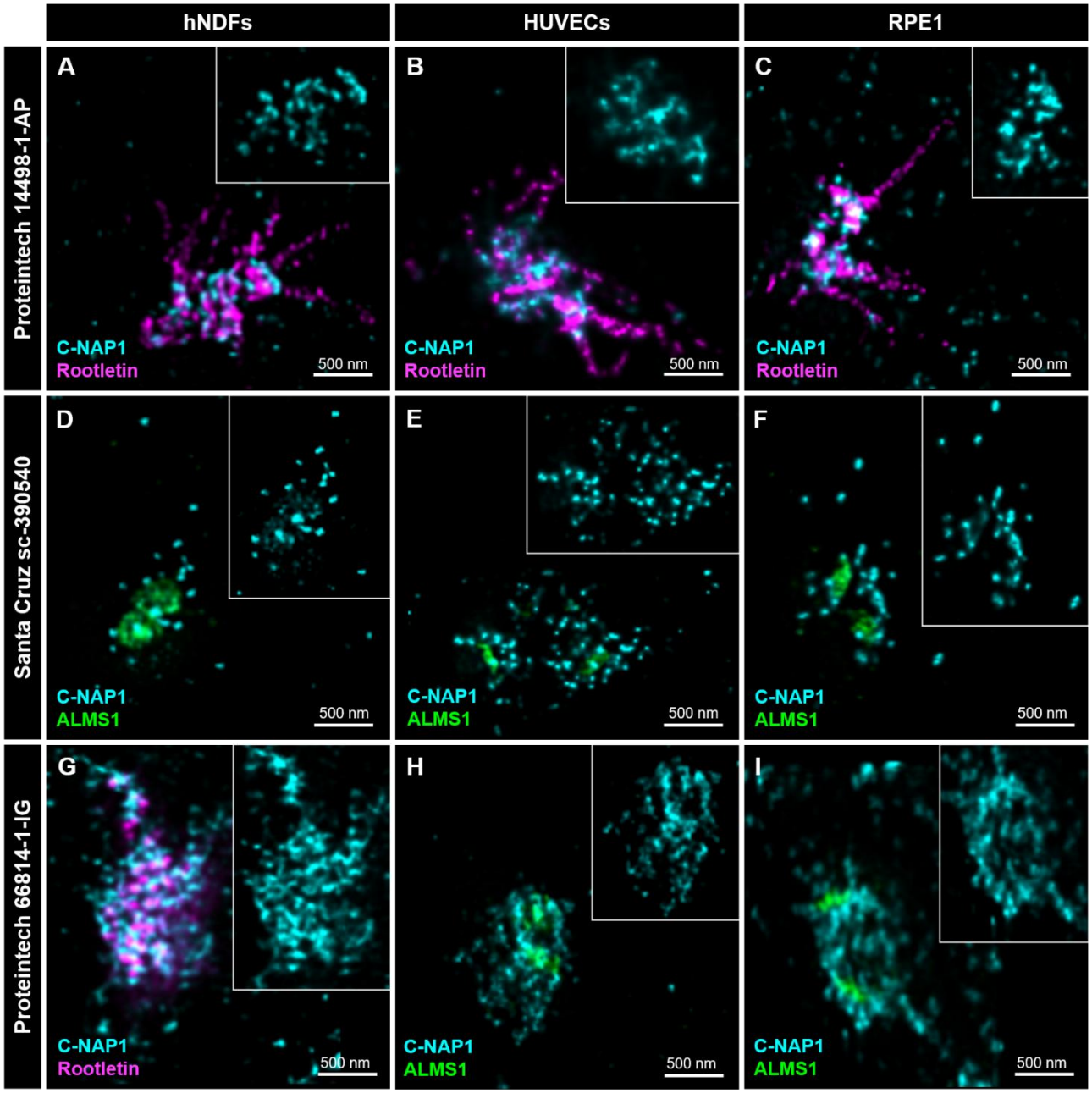
Supplementary Figures



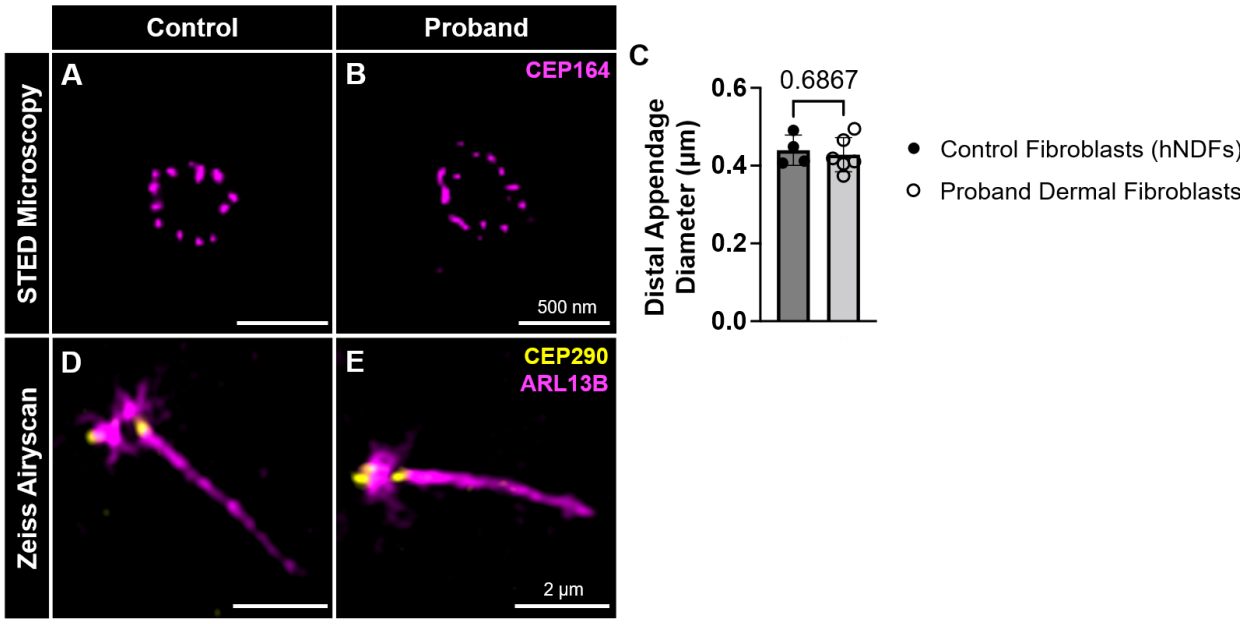
Supplementary Figure 1. Scree plot of principal component analysis showing the majority of variance explained by PC1



Supplementary Figure 2. Normalized counts of genes of interest categorized by A) fibroblast activation, B) ECM, C) EndoMT, and D) cilia.



Supplemental Figure 3. C-NAP1 is a discontinuous structure that is concentrated near the centriole proximal end and can be visualized within Rootletin fibers.
(**A-I**) STED super-resolution microscopy of C-NAP1 in three unique cell types (hNDFs, HUVECs, RPE1) with three independent antibodies (Proteintech 14498-1-AP, Santa Cruz sc-390540, Proteintech 66814-1-IG), co-stained with either ALMS1 (**D,E,F,H,I**) or Rootletin (**A,B,C,G**). Regardless of the cell type or antibody used, C-NAP1 can be seen as scattered, punctate, and associated with Rootletin fibers. Scale bars: 500 nm.



Supplemental Figure 4. ALMS1 loss does not affect the distal structures of the centriole. (A,B) STED super-resolution imaging of the distal appendages (CEP164) in control (**A**; hNDFs) and (**B**; proband fibroblasts). **(C)** Quantification of distal appendage diameters in control and proband cells. Centriole distal appendages are intact with unchanged diameters in proband cells. **(D,E)** Zeiss 880 Airyscan imaging of the transition zone (CEP290) in control and proband fibroblasts. CEP290 is visible and accurately localized to the base of the axoneme in control and proband cells. Statistics for panel C were calculated using an unpaired t test. Each data point represents an independent experiment. Data are presented as the mean ± S.D. Scale bars: 500 nm (**A,B**), 2 µm (D,E).