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Supplemental Data

Exome Sequencing and Functional Analysis

Identifies *BANF1* Mutation as the Cause

of a Hereditary Progeroid Syndrome

Xose S. Puente, Victor Quesada, Fernando G. Osorio, Rubén Cabanillas, Juan Cadiñanos, Julia M. Fraile, Gonzalo R. Ordóñez, Diana A. Puente, Ana Gutiérrez-Fernández, Miriam Fanjul-Fernández, Nicolas Lévy, José M. P. Freije, and Carlos López-Otín



Figure S1. Clinical Characteristics of Atypical Progeria in Patient II-1 from Family A

Frontal and dorsal view of the patient, and detailed pictures of head, ears, feet and hands, illustrating progeroid features and severe skeletal abnormalities.



Figure S2. Clinical Characteristics of Atypical Progeria in Patient II-1 from Family B

Frontal and dorsal view of the patient, and detailed pictures of head, ears, feet and hands, illustrating progeroid features and severe skeletal abnormalities.

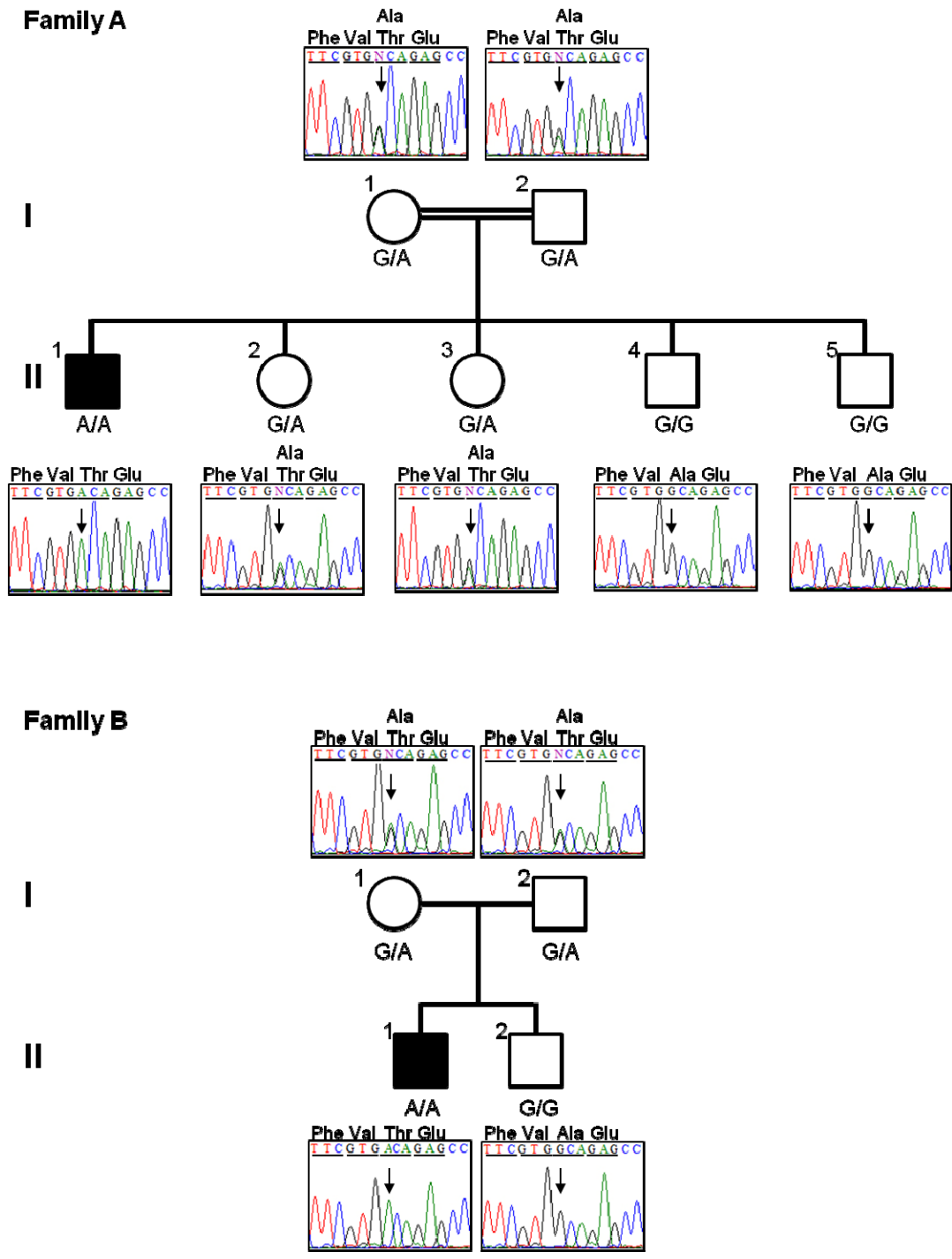


Figure S3. Pedigrees and Electropherograms Corresponding to the p.Ala12Thr Mutation in BAF

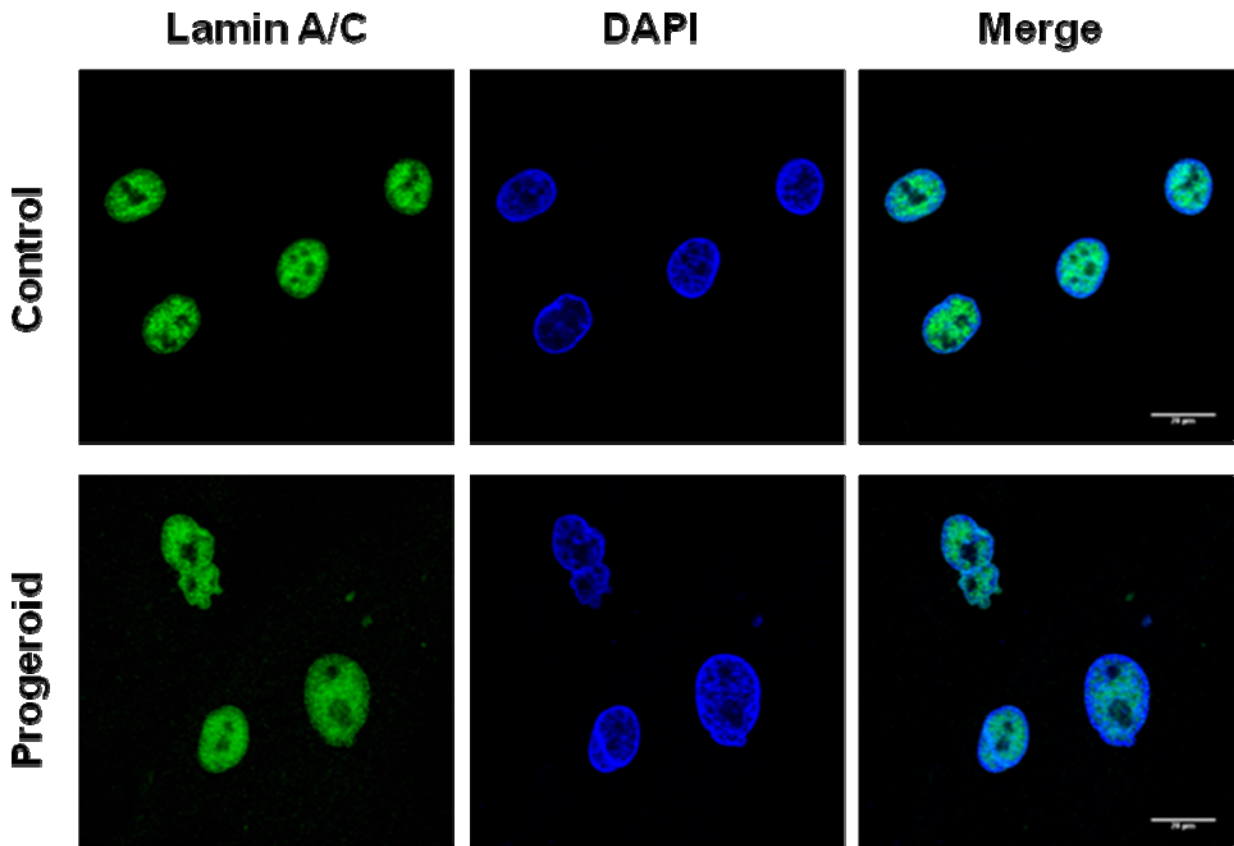


Figure S4. Lamin A/C in Fibroblasts Homozygous for the p.Ala12Thr Mutation in BAF

Lamin A/C distribution was analyzed by immunofluorescence and confocal microscopy in primary fibroblasts from the progeroid patient II-1 (family A) as well as in control fibroblasts.