SUPPLEMENTAL INFORMATION

COL6A2 Recessive C-Globular Missense Mutations in Ullrich Congenital Muscular Dystrophy: Role of the C2a Splice Variant

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Figure 1S. Immunoblot analysis of fibroblasts from a healthy donor with the α2(VI) collagen chain specific polyclonal antibody. Human dermal fibroblasts were grown until 1 day after confluency, and then switched to serum-free medium for 24 hr. Cell lysate (30 μg total protein/lane) and a 100 μl aliquot of the serum-free culture media precipitated with 0.9 ml of 100% EtOH were separated on a 3-8% polyacrylamide gel. The positions of the sample wells and protein size markers are indicated. The antisera detected a major band of approximately 150 kDa in both culture medium and cell lysate and a minor band smaller than the α2(VI) collagen in the cell lysate (arrow).

Figure 2S. Molecular modeling analysis of the COL6A2 C1 and C2 subdomains and the missense mutations. *A* and *B*. Schematic drawings of the C1 (A) and C2 (B) subdomains based on the three dimensional structure of the vWF-A1 domain. The locations of the E624K and R876S mutations, the previously reported L837P and N876 deletion mutations, and the N- and C-termini of the subdomains are shown. *C* and *D*. The effects of E624K and R876S mutations on the electrostatic potential (EP) of the protein. Computer analyses were performed with the regions from amino acids D615 to D651 in the C1 subdomain (C) and from P785 to S841 in the C2 subdomain (D). Red color: highly positive EP; Blue color: highly negative EP. The two missense mutations significantly alter the EP of the adjacent regions.

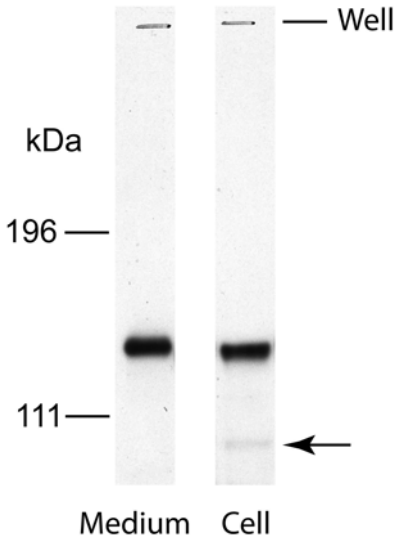

Figure 1S

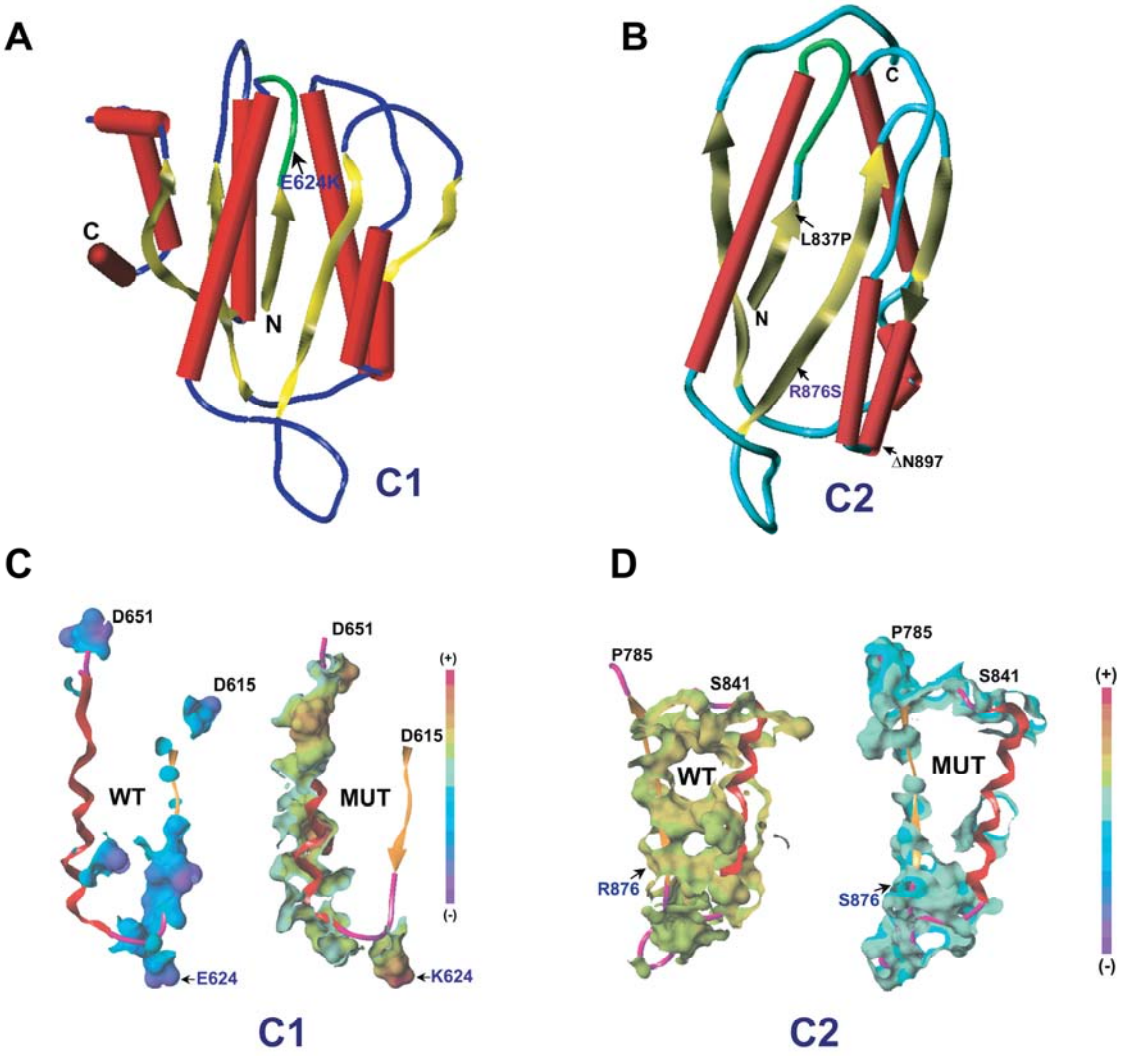
A
E624K
C
N
C1
B
C
L837P
N
R876S
ΔN897
C2
C
D651
D615
WT
E624
D651
D615
MUT
K624
(+)
(-)
C1
D
P785
S841
WT
R876
P785
S841
MUT
S876
(+)
(-)
C2

Figure 2S