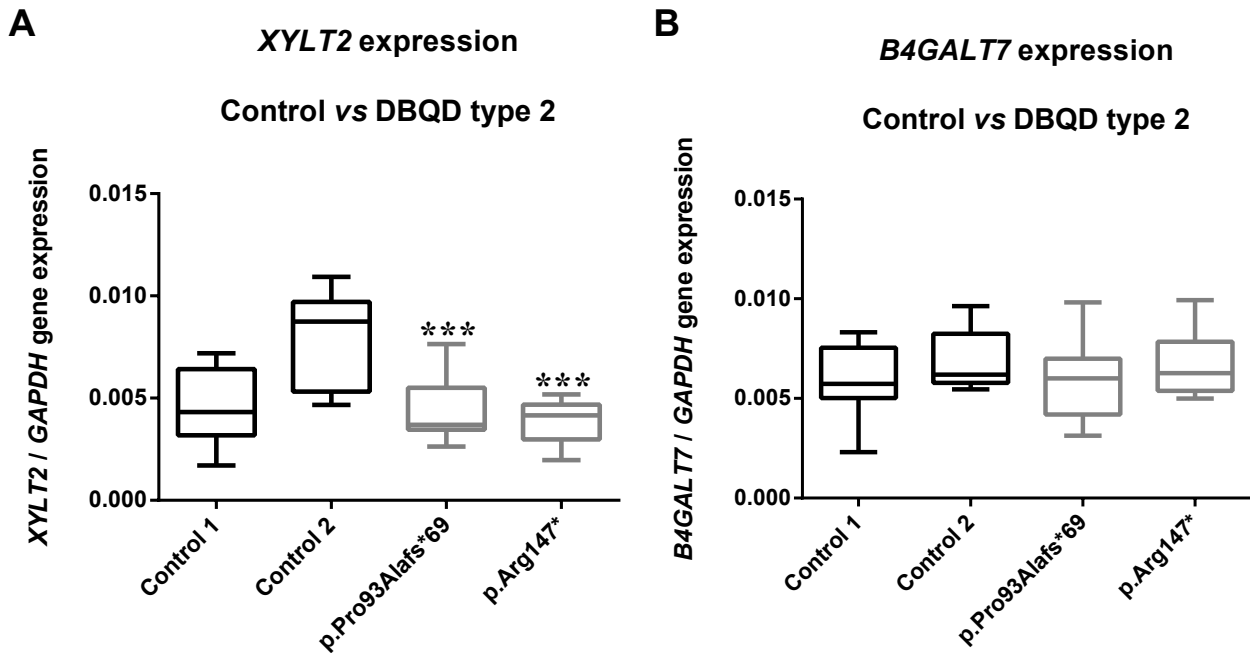


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Supplemental Data

## ***XYLT1* Mutations in Desbuquois Dysplasia Type 2**

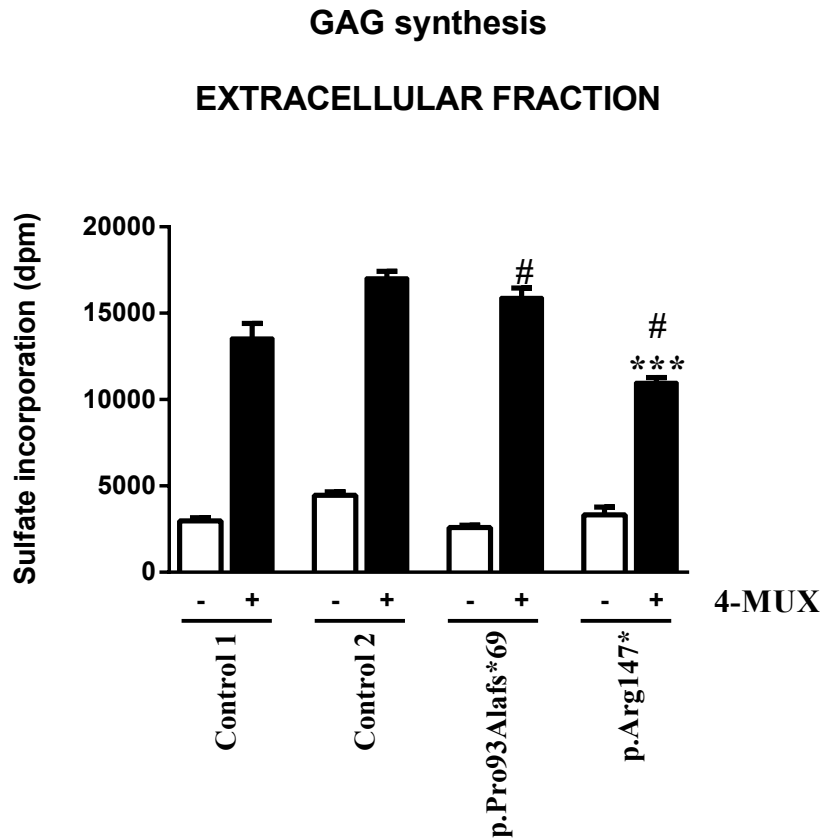
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**Figure S1. cDNA level of *XYLT2* and *B4GALT7* in controls and two cultured individual fibroblasts (p.Arg147\* and p.Pro93Alafs\*69)**

cDNA levels were normalized to *GAPDH*. Individuals p.Arg147\* and p.Pro93Alafs\*69 are female aged 14 and male aged 23 respectively. Control 1 (female) and 2 (male) are aged 2.5 and 12 respectively. Control (black boxes) and individual (grey boxes) data are presented as box plots and are the mean of 3 independent experiments performed in quadruplicate. \*\*\* $p < 0.001$  compared to control 2 using 1-way ANOVA with Tukey's *post hoc* test.

Note that mutations in *XYLT1* affect cDNA level of *XYLT2* (A) but not *B4GALT7* (B)



**Figure S2. Sulfate incorporation of extracellular fractions in controls and the two cultured individual fibroblasts in the presence of 4-MUX (5  $\mu$ M) or vehicle (DMSO)**

The black (+4-MUX) and white (DMSO) bars represent the mean  $\pm$  SEM of 2 independent experiments performed in triplicate. Control 1 (female) and 2 (male) are aged 2.5 and 12 respectively. # $p < 0.05$  when compared to control 1, \*\*\* $p < 0.001$  when compared to control 2 using 1-way ANOVA with Tukey's *post hoc* test.

Note that the biosynthesis of secreted PGs is enhanced by the 4-MUX treatment in the cultured fibroblasts but at variable level