Supplemental information

A pseudoautosomal glycosylation disorder prompts the revision of dolichol biosynthesis

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Supplemental Table S3

Table S3. Summary of patient-derived cell lines used in this study, related to Table S2 and Figures 1, 2, 3, 6, S1, S2, and S3.

Cell type	Identifier	Variant 1	Variant 2	Literature
EBV-immortalized lymphoblastoid cells	Control 1	-	-	-
	Control 2	-	-	-
	Patient 1	c.541G>T; p.(Val181Phe) (X)	c.541G>T; p.(Val181Phe) (X)	This study
	Patient 3	c.541G>T; p.(Val181Phe) (X)	c.643C>T; p.(Leu215Phe) (Y)	This study
	Patient 4	c.541G>T; p.(Val181Phe) (X)	c.643C>T; p.(Leu215Phe) (Y)	This study
	P3/P4 mother	c.541G>T; p.(Val181Phe) (X)	-	This study
	P3/P4 father	-	c.643C>T; p.(Leu215Phe) (Y)	This study
	SRD5A3-CDG	c.436G>A, p.(Glu146Lys)	c.436G>A, p.(Glu146Lys)	Kousal *et al.* 2019 [S1]
Dermal fibroblasts	Control 1	-	-	-
	Control 2	-	-	-
	Patient 1 (DHRSX/Y-CDG)	c.541G>T; p.(Val181Phe) (X)	c.541G>T; p.(Val181Phe) (X)	This study
	Patient 3 (DHRSX/Y-CDG)	c.541G>T; p.(Val181Phe) (X)	c.643C>T; p.(Leu215Phe) (Y)	This study
	SRD5A3-CDG 1	c.57G>A; p.(Trp19*)	c.224_225insA; p.(Tyr75*)	-
	SRD5A3-CDG 2	c.320G>A; p.(Trp107*)	c.320G>A; p.(Trp107*)	Rymen *et al.* 2012 [S2]
	SRD5A3-CDG 3	c.530_531delTG; p.(Val177Alafs*42)	c.530_531delTG; p.(Val177Alafs*42)	Rymen *et al.* 2012 [S2]
	SRD5A3-CDG 4	c.79_80insGGAA; p.(Leu27Argfs*45)	c.79_80insGGAA; p.(Leu27Argfs*45)	Rymen *et al.* 2012 [S2]
	SRD5A3-CDG 5	c.634delT; p.(Trp212Glyfs*46)	c.634delT; p.(Trp212Glyfs*46)	Rymen *et al.* 2012 [S2]
	SRD5A3-CDG 6	c.57G>A; p.(Trp19*)	c.57G>A; p.(Trp19*)	Rymen *et al.* 2012 [S2]

(X) = variant present on the X chromosome; (Y) = variant present on the Y chromosome

Supplementary references:

[S1] Kousal, B., Honzík, T., Hansíková, H., Ondrušková, N., Čechová, A., Tesařová, M., Stránecký, V., Meliška, M., Michaelides, M., & Lišková, P. (2019). Review of SRD5A3 Disease-Causing Sequence Variants and Ocular Findings in Steroid 5α-Reductase Type 3 Congenital Disorder of Glycosylation, and a Detailed New Case. *Folia biologica*, *65*(3), 134–141.

[S2] Rymen, D., Keldermans, L., Race, V., Quelhas, D., Snyder, F., Alehan, F., Erol, I., Mübeccel, D., Gokçay, G., Mitchell, G., Matthijs, G., & Jaeken, J. (2012). *SRD5A3-CDG: an important group of dolichol-phosphate synthesis defects* (Vol. 35, pp. S115–S115). Proceedings of the SSIEM 2012.