Supplementary Material to "Cockayne Syndrome: The many challenges and approaches to understand a multifaceted disease"

Table S1 - Homozygous and heterozygous *ERCC6 (CSB*) mutations and their effects on patients phenotype.

***CSB* - Homozygous**			
Mutation	**Patient code**	**CS type**	**Reference**
p.Arg735*	CS17LO CS128ST	CS I	(Calmels *et al.*, 2018)
p.Arg652*	CS10X	?	(Calmels *et al.*, 2018)
p.(Gln854*)	CS144ST	CS II	(Calmels *et al.*, 2018)
p.(Asp1355Valfs*32)	CS071ST CS222ST	CS I	(Calmels *et al.*, 2018)
	CS204ST CS221ST	CS III	(Calmels *et al.*, 2018)
p.Leu72Cysfs*12	CS18BR	CS I	(Calmels *et al.*, 2018)
p.(Glu214*)	CS010ST	CS I	(Calmels *et al.*, 2018)
p.Ser357*	CS13PV	CS I	(Calmels *et al.*, 2018)
p.(Thr377Glnfs*28)	CS201ST	CS II	(Calmels *et al.*, 2018)
p.Lys478Thrfs*9	CS19PV	CS II	(Calmels *et al.*, 2018)
p.Trp517*	CS4TAN CS8TAN	CS II	(Calmels *et al.*, 2018)
p.(Glu564*)	CS107ST	CS II	(Calmels *et al.*, 2018)
p.(*Asp646Asn*)	CS253ST	CS II	(Calmels *et al.*, 2018)
p.Arg652*	CS13MA	CS II	(Calmels *et al.*, 2018)
p.(Thr659Cysfs*24)	CS232ST	CS II	(Calmels *et al.*, 2018)
p.Phe665Tyrfs*18	CS1PL	CS III	(Calmels *et al.*, 2018)

p.Gly715*	CS23PV	CS I	(Calmels *et al.*, 2018)
p.Phe665_Gln723del	CS11MA	CS II	(Calmels *et al.*, 2018)
p.Asn760Lysfs*2	CS8PV	CS II	(Calmels *et al.*, 2018)
p.Met867Thrfs*14	CS31PV	CS II	(Calmels *et al.*, 2018)
p.Leu875Pro	CS20PV	CS II	(Calmels *et al.*, 2018)
p.Ala944Thrfs*10	CS11LO	CS II	(Calmels *et al.*, 2018)
p.Gln976Trpfs*70	CS23BR	CS II	(Calmels *et al.*, 2018)
p.(Thr1018Asnfs*32)	CS210ST	CS II	(Calmels *et al.*, 2018)
p.[Gln156*; Ser142Asnfs*4]	CS14LO	CS I	(Calmels *et al.*, 2018)
p.[Arg176*;Ser142Asnfs*4]	CS27PV	CS I	(Calmels *et al.*, 2018)
p.(Ser429Lysfs*7)	CS10LO	CS I	(Calmels *et al.*, 2018)
p.Val161Serfs*5	CS1BR	?	(Calmels *et al.*, 2018)
p.(Tyr1179Leufs*22)	CS241ST	?	(Calmels *et al.*, 2018)
p.[Gln156*; Ser142Asnfs*4]	CS3SH	?	(Calmels *et al.*, 2018)
p.Arg670Trp	?	CS II	(Chebly *et al.*, 2018)
p.Arg453*	CS1PV CS3PV	CS II	(Colella *et al.*, 1999)
p.Arg735*	GM10905 GM10903	DSC	(Colella *et al.*, 2000)
p.Lys345Asnfs*24	CSB1NH	CS II	(Falik-Zaccai *et al.*, 2008)
p.Arg77Ilefs*6	KPSX6	CS III	(Hashimoto *et al.*, 2008)
p.Arg77*	UVS1KO	UVSS	(Horibata *et al.*, 2004)
p.Arg1288*	XVI-1 XVI-2 XIII-6 XIII-7 XIV-2 XIV-6	COFS	(Jaakkola *et al.*, 2010)

p.Lys971Tryfs*14	?	CS II	(Kou *et al.*, 2018)
p.Arg683*	CS789VI	COFS	(Laugel *et al.*, 2008)
p.Val763_Gln794del	CS514VI CS385VI	CS II	(Laugel *et al.*, 2009)
p.Arg652*	08TR4	CS II	(Laugel *et al.*, 2009)
p.Arg1288*	CS817VI	CII	(Laugel *et al.*, 2009)
p.Arg1087*	CS6BI CS8BI	CS II	(Laugel *et al.*, 2009)
p.Gln854*	CS9MA	CS II	(Laugel *et al.*, 2009)
p.Glu218Glyfs*4	AENT4	CS I	(Laugel *et al.*, 2009)
p.Asp1355Valfs*32	CS1SACT	CS I	(Laugel *et al.*, 2009)
p.Glu182Asnfs*4	CS799VI	CS III	(Laugel *et al.*, 2009)
p.Gly715*	08STR1	CS II	(Laugel *et al.*, 2009)
p.[Arg176*;Gly399Asp; Ser1321Cys]	?	CS III	(Luo *et al.*, 2014)
p.Trp517*	CS2TAN	CS I	(Mallery *et al.*, 1998)
p.Arg735*	CS1TAN	CS I	(Mallery *et al.*, 1998)
p.Trp851Arg	CS3TAN	CS I	(Mallery *et al.*, 1998)
p.Val957Gly	CS1IAF	CS I	(Mallery *et al.*, 1998)
p.Lys1239Glufs*2	?	COFS	(Meira *et al.*, 2000)
p.Arg637Serfs*34	?	CS III	(Swartz *et al.*, 2015)
p.Pro934Thr	?	CS I	(Wilson *et al.*, 2016)
p.Trp936Cys	?	CS II	(Wilson *et al.*, 2016)
p.Gln463*	?	CS I	(Zhang *et al.*, 2011)
***CSB* - Heterozygous**			
Mutation	**Patient code**	**CS type**	**Reference**

p.*Ser687Leu* p.Arg1288*	CS278ST	CS II	(Calmels *et al.*, 2016)
p.[Gln156;Ser142Asnfs*4] p.Leu471Glnfs*16	CS1SH	CS I	(Calmels *et al.*, 2018)
p.[Arg176*;Ser142Asnfs*4] p.Arg652*	CS1SO	CS I	(Calmels *et al.*, 2018)
p.[Arg176*;Ser142Asnfs*4] *p.Trp589del*	CS1GO	CS I	(Calmels *et al.*, 2018)
p.Ser142Asnfs*4 p.Arg735*	CS26PV	CS III	(Calmels *et al.*, 2018)
p.Arg453* p.Val763Ilefs*7	CS1WR	CS II	(Calmels *et al.*, 2018)
p.Arg467_Arg562del p.Arg1318Glyfs*12	CS058ST	CS I	(Calmels *et al.*, 2018)
p.Tyr510_Arg562del p.Glu608_Gln723del	CS18PV	CS II	(Calmels *et al.*, 2018)
p.Phe563Argfs*3 p.[Gln943*; Gln943Profs*8; Asp904_Gln943del]	CS17PV	CS I/II	(Calmels *et al.*, 2018)
p.0 p.Phe665_Gln723del	CS10MA CS1LI	CS II	(Calmels *et al.*, 2018)
	CS2BL	?	(Calmels *et al.*, 2018)
p.*Arg670Trp* p.Val763_Gln794del	CS18LO	CS II	(Calmels *et al.*, 2018)
p.Arg683* p.Phe665_Gln723del	CS12MA	CS II	(Calmels *et al.*, 2018)
p.*Ser687Leu* p.Arg735*	CS1GGO	CS II	(Calmels *et al.*, 2018)
p.Leu700Valfs*60 p.Arg735*	CS22PV CS28PV	CS I	(Calmels *et al.*, 2018)
p.Phe665_Gln723del p.Arg1221*	CS8MA	CS II	(Calmels *et al.*, 2018)
p.Phe665_Gln723del p.Arg1087*	CS19LO	CS II	(Calmels *et al.*, 2018)

p.Arg735* p.Lys1203fs	CS2GR	CS I	(Calmels *et al.*, 2018)
p.*Trp851Arg* p.Arg1318Glyfs*12	CS21PV	CS I	(Calmels *et al.*, 2018)
p.0 p.Arg612*	CS195ST	CS II	(Calmels *et al.*, 2018)
p.[Gln156*;Ser142Asnfs*4] p.Arg947*	CS5MA	?	(Calmels *et al.*, 2018)
p.[Gln156*;Ser142Asnfs*4] p.Arg735*	CS2LE	III?	(Calmels *et al.*, 2018)
p.Phe563Argfs*3 p.Arg735*	CS25PV	?	(Calmels *et al.*, 2018)
p.Arg683* p.Arg1288*	CS32LO	?	(Calmels *et al.*, 2018)
p.Phe665_Gln723del p.*Gln953Lys*	CS3BL	?	(Calmels *et al.*, 2018)
p.Arg735* p.0	CS14PV	CS I	(Calmels *et al.*, 2018)
p.Ala944Thrfs*10 p.Tyr1179Leufs*22	CS1BEL	?	(Calmels *et al.*, 2018)
p.Arg612* p.Arg975Trp	?	CS II	(He *et al.*, 2017)
p.Arg857* p.Arg1087*	CS816VI	COFS	(Laugel *et al.*, 2008)
p.Leu871Pro p.Lys1172*	CS881VI	COFS	(Laugel *et al.*, 2009)
p.Leu987Pro p.Met752_Gln762del	CS797VI	COFS	(Laugel *et al.*, 2009)
p.Val724_Gln762del p.Trp686Cys	CS683VI	CS II	(Laugel *et al.*, 2009)
p.Gln723* p.Leu860del	CS3LE	CS II	(Laugel *et al.*, 2009)
p.Gly715* p.Arg857*	08STR1	CS II	(Laugel *et al.*, 2009)

p.Val724_Gln762del p.Ala944Thrfs*10	CS1GL	CS II	(Laugel *et al.*, 2009)
p.Met867Thrfs*14 p.Lys1198Argfs*4	CS117VI	CS II	(Laugel *et al.*, 2009)
p.Gln956Argfs*7 p.Ser687Leu	CS360VI	CS II	(Laugel *et al.*, 2009)
p.Val417Serfs*7 p.0	CS179VI	CS II	(Laugel *et al.*, 2009)
p.Gln156* p.His1263Glnfs*67	CS21BR	CS I	(Laugel *et al.*, 2009)
p.Trp236* p.Pro500Glnfs*43	CS493VI	CS I	(Laugel *et al.*, 2009)
p.Arg68Profs*13 p.Asn680Asp	CS784VI	CS I	(Laugel *et al.*, 2009)
p.Arg637Serfs*34 p.Asp749Glufs*4	CS543VI	CS III	(Laugel *et al.*, 2009)
p.Glu182Asnfs*4 p.Glu379*	CS393VI	CS III	(Laugel *et al.*, 2009)
p.Ser1240_Val1260delinsIle p.Arg735*	CS823VI	CS III	(Laugel *et al.*, 2009)
p.Lys1203fs p.Met867Thrfs*14	CS1ABR	CS II	(Mallery *et al.*, 1998)
p.Tyr1179Ilefs*22 p.Phe665_Gln723del	CS2BE	?	(Mallery *et al.*, 1998)
p.Arg735* p.Arg453*	25627	CS I	(Mallery *et al.*, 1998)
p.Gln184* p.Arg670Trp	CS4BR	CS I	(Mallery *et al.*, 1998)
p.Arg670Trp p.Pro1042Leu	CS2BI	CS I	(Mallery *et al.*, 1998)
p.Ser142Asnfs*4	?	?	(Shehata *et al.*, 2014)
p.Lys337* p.Tyr834Cysfs*25	CS1AN	CS I	(Troelstra *et al.*, 1992)

p.(Glu215*) p.(Ser1259*)	?	?	(Wu *et al.*, 2016)
p.Asp532Gly p.Leu536Trp	?	CS I	(Yu *et al.*, 2014)

- Question mark (?) is used to refer to missing informations and uncertainties.
- The mutations nomenclature was described following the HGVS recommendations (Dunnen *et al.*, 2006).
- * is used to indicate a stop codon.

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