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

**Table S2 -** Homozygous and heterozygous *ERCC8 (CSA*) mutations and their effects on patients phenotype.

| *CSA* - Homozygous | | | |
|---|---|---|---|
| Mutation | Patient code | CS type | Reference |
| p.Asp266Gly | 3_1<br>3_2 | CS I | (Bertola *et al.*, 2006) |
| p.Glu55Lysfs*13 | CS30PV | CS I | (Biancheri 2014) |
| p.(*His244Tyr*) | CS276ST | CS III ? | (Calmels *et al.*, 2016) |
| p.*Thr265Pro* | CS260ST | CS III ? | (Calmels *et al.*, 2016) |
| p.Glu13* | CS15PV<br>CS2JE | CS I | (Calmels *et al.*, 2018) |
| p.(Arg268*) | CS240ST | CS I/II | (Calmels *et al.*, 2018) |
| p.*Leu271Pro* | CS3BR | CS II | (Calmels *et al.*, 2018) |
| p.[Tyr322*;Val282_Gln347del; Val282_Glu374del] | CS9IAF | CS I | (Calmels *et al.*, 2018) |
| p.Val27_Arg92del | CS1LE | CS I | (Calmels *et al.*, 2018) |
| p.Leu251Tyrfs*18 | CS172ST | CS II | (Calmels *et al.*, 2018) |
| p.Ala240Glyfs*8 | CS7PV | CS II | (Calmels *et al.*, 2018) |
| p.Tyr200Lysfs*12 | CS1GLO | CS I | (Calmels *et al.*, 2018) |
| p.Asn75Glnfs*9 | CS4PV | CS II | (Calmels *et al.*, 2018) |
| p.(Gln106*) | CS263ST | CS I | (Calmels *et al.*, 2018) |
| p.Thr134Leufs*13 | CS261ST | CS I/II | (Calmels *et al.*, 2018) |
| p.Tyr322* | ? | CS I | (Chebly *et al.*, 2018) |

| | | | |
|---|---|---|---|
| p.*Tyr350Cys* | GM02965 | CS III | (Kennedy *et al.*, 1980) |
| p.Gly184Aspfs28* | CSA10NO | CS I | (Kleppa *et al.*, 2007) |
| p.Asp266Gly | 08STR2 | CS I | (Laugel *et al.*, 2009) |
| p.Ala207_Ser209del | CS794VI | CS I | (Laugel *et al.*, 2009) |
| p.Leu202Ser | 08STR3 | CS I | (Laugel *et al.*, 2009) |
| p.Ala160Thr | CS852VI | CS III | (Laugel *et al.*, 2009) |
| p.Gly184Aspfs28* | CS7BI<br>CS7BI2<br>CS7BI3 | CS I | (Laugel *et al.*, 2009) |
| p.Tyr322* | CS886VI<br>CS887VI | CS I | (Laugel *et al.*, 2009) |
| p.Tyr200Lysfs*12 | CS24PV<br>CS417VI | ? | (Laugel *et al.*, 2009) |
| p.Glu13* | CS218ST | CS I | (Laugel *et al.*, 2013) |
| p.Trp361Cys | UVS$^{S}$VI | UVSS | (Nardo *et al.*, 2009) |
| p.Thr134Leufs*13 | CS133NY | CSI/III | (Rapin *et al.*, 2009) |
| p.Glu374Asp | ? | CS I | (Taghdihi 2017) |
| ***CSA* - Heterozygous** | | | |
| Mutation | Patient code | CS type | Reference |
| p.Val105Thrfs*6<br>p.Thr204Lys | ? | CS I | (Bertola *et al.*, 2006) |
| p.Glu13*<br>p.Ala205Pro | AGO7075 | CSI | (Cao *et al.*, 2004) |
| p.Thr134Leufs*13<br>p.Thr134Valfs*7 | CS6PV | CS I | (Calmels *et al.*, 2018) |
| p.Asp199Metfs*14<br>p.Ser220* | CS16PV | CS I | (Calmels *et al.*, 2018) |
| *p.Ala207_Ser209del*<br>p.0 | CS1JE | CS I | (Calmels *et al.*, 2018) |

| | | | |
|---|---|---|---|
| p.Pro95Leufs*30<br>p.Val161Serfs*5 | CS9LO | CS<br>I/II | (Calmels *et al.*, 2018) |
| *p.Ser119Leu*<br>p.Ala207_Ser209del | CS040ST | CS I | (Calmels *et al.*, 2016) |
| p.Tyr100*<br>p.Thr134Valfs*7 | CS11PV | CS I | (Conte *et al.*, 2009) |
| p.Leu132fs*137<br>p.Ala240fs*243 | ? | CS I | (Gu *et al.*, 2017) |
| p.Trp194Cys<br>p.Thr24Asnfs*11 | CS655VI | CS I | (Laugel *et al.*, 2009) |
| p.Asp266Gly<br>p.Ala240Glyfs*8 | CS861VI | CS I | (Laugel *et al.*, 2009) |
| p.Asp93Leufs*26<br>p.Gln106Pro | CS2SE | ? | (Ren *et al.*, 2003) |
| p.Ala160Val<br>p.Glu13* | CS3BE | CS I | (Ridley *et al.*, 2005) |
| p.Ala207_Ser209<br>p.Asp93Leufs*26 | ? | CS I | (Xie *et al.*, 2017) |

- 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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