|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| Table e-5. Genotype-Phenotype Correlations | | | | | | |
| Gene |  | **DNA** | **Protein** | **Type** | **Patient Class** | **References** |
| GYPH |  | - | p.V22fs | Frameshift | 1 | (Reiss *et al.*, 2001) |
| GYPH |  | c.1739A>C | p.D580A | Missense | 1 | (Reiss *et al.*, 2011) |
| MOCS1A |  | c.217C>T | p.R73W | Missense | 1 | (Gümüş *et al.*, 2010) |
| MOCS1A |  | c.251\_418del | p.C84fs | Frameshift | 1 | (Carmi-Nawi *et al.*, 2011) |
| MOCS1A |  | c.253C > T | p.Q85X | Nonsense | 1 | (Zaki *et al.*, 2016) |
| MOCS1A |  | c.377G>A | p.G126D | Missense | 1 | (Mills *et al.*, 2012) |
| MOCS1A |  | c.418+1G>A | Skipping exon 2 | Splice site | 1 | (Reiss *et al.*, 1999) |
| MOCS1A |  | c.664\_666dupCGA | p.R222\_G223insR | Insertion (in frame) | 1/2 | (Sass *et al.*, 2010) Class 1  (Ichida *et al.*, 2006) Class 2 |
| MOCS1A |  | c.722delT | p.L241RfsX6 | Frameshift/Truncation | 1 | (Zaki *et al.*, 2016) |
| MOCS1A |  | c.955C>T | p.R319X | Nonsense | 1 | (Mills *et al.*, 2012) |
| MOCS1A |  | c.971G>A | p.G324E | Missense | 1 | (Zaki *et al.*, 2016) |
| MOCS1A |  | c.1064T>C | p.I355T | Missense | 1 | (Macaya *et al.*, 2005) |
| MOCS1A |  | c.1102+1G>A | Skipping exon 8 | Splice site | 1 | (Zaki *et al.*, 2016) |
| MOCS1A |  | c.1165+6T>C | Skipping exon 9 | Splice site | 2 | (Arenas *et al.*, 2009) |
| MOCS1AB |  | c.1643C>A | p.A543E | Missense | 1 | (Higuchi *et al.*, 2014) |
| MOCS1AB |  | c.1777G>A | p.G588R | Missense | 1 | (Veldman *et al.*, 2010) |
| MOCS2A |  | c.3G>A | p.M1I | Missense | 2 | (Zaki *et al.*, 2016)(Megahed *et al.,* 2016) |
| MOCS2A |  | c.16C>T | p.Q6X | Nonsense | 2 | (Johnson *et al.*, 2001) |
| MOCS2A |  | c.19G>T | p.V7F | Missense | 2 | (Johnson *et al.*, 2001) |
| MOCS2A |  | c.130C>T | p.R44X | Nonsense | 1 | (Per *et al.*, 2007) |
| MOCS2A |  | c.220C>T | p.Q74X | Nonsense | 1 | (Sie *et al.*, 2010)(Mills *et al.*, 2012) |
| MOCS2A |  | c.265T>C | p.X89Q | Nonsense | 1 | (Kikuchi *et al.*, 2012) |
| MOCS2A |  | c.266A>G | p.X89W | Nonsense | 1 | (Kikuchi *et al.*, 2012) |
| MOCS2A/B |  | c.252\_253insC | MOCS2A:I85fs; MOCS2B: p.L23fs | Frameshift | 1 | (Nagappa *et al.*, 2015) |
| MOCS2B |  | c.226 G>A | p.G76R | Missense | 1 | (Mills *et al.*, 2012) |
| MOCS2A |  | c.501+ 2delT |  | Splice site | 1 | (Edwards *et al.*, 2015) |
| MOCS2B |  | c.564G>C | p.G126A | Missense | 2 | (Alkufri *et al.*, 2013) |
| MOCS2B |  | c.564+1G>A | skipping exon 5 | Splice site | 1 | (Struys *et al.*, 2012)(Stence *et al.*, 2013) |
| MOCS2B |  | c.726\_727delAA | p.K180fs | Frame shift | 2 | (Alkufri *et al.*, 2013) |
| MOCS3 |  | c.769G >A | p.A257T | Missense | 2 | (Huijmans *et al.*, 2017) |
| SUOX |  | c.794C>A | p.A265D | Missense | 1 | (Edwards *et al.*, 1999) |
| SUOX |  | Not reported | p.C508Y | Missense | 1 | (Bindu *et al.*, 2011) |
| SUOX |  | c.520delG | p.D174TfsX12 | Frameshift/Truncation | 1 | (Seidahmed *et al.*, 2005) |
| SUOX |  | c.884G>A | p.G295E | Missense | 1 | (Zaki *et al.*, 2016) |
| SUOX |  | c.1355G>A | p.G452D | Missense | 1 | (Chen *et al.*, 2014) |
| SUOX |  | c.427C>A | p.H143N | Missense | 2 | (Del Rizzo *et al.*, 2013)(Mills *et al.*, 2012) |
| SUOX |  | c.1136A>G | p.K379R | Missense | 1 | (Sass *et al.*, 2010)(Holder *et al.*, 2014) |
| SUOX |  | c.182T>C | p.L61P | Missense | 2 | (Rocha *et al.*, 2014) |
| SUOX |  | c.571\_574delCAGC | p.Q191GfsX11 | Frameshift/Truncation | 1 | (Johnson *et al.*, 2002) |
| SUOX |  | c.571delC | p.Q191SfsX12 | Frameshift/Truncation | 1 | (Rupar *et al.*, 1996) |
| SUOX |  | c.650G>A | p.R217Q | Missense | 2 | (Lam *et al.*, 2002)(Garrett *et al.*, 1998) |
| SUOX |  | c.1585C>T | p.R529X | Nonsense | 1 | (Hoffmann *et al.*, 2007) |
| SUOX |  | c.1280C>A | p.S427Y | Missense | 1 | (Edwards *et al.*, 1999) |
| SUOX |  | c.1406\_1421delCCTGGCAGGTGGCTAA | p.T469SfsX19 | Frameshift/Truncation | 1 | (Basheer *et al.*, 2007) |
| SUOX |  | c.1100\_1101delTG | p.V367GfsX47 | Frameshift/Truncation | 1 | (Hoffmann *et al.*, 2007) |
| SUOX |  | c.1313\_1316delTAGA | p.V438DfsX4 | Frameshift/Truncation | 1 | (Tan *et al.*, 2005) |
| SUOX |  | 1200C>G | Y400X | Nonsense | 1 | (Cho *et al.*, 2013)(Chen *et al.*, 2014)(Lee *et al.*, 2017) (Basheer *et al.*, 2007) |
| SUOX |  | -137+2dup |  | Splice site | 1 | (Bindu *et al.*, 2011) |
| SUOX |  | 228+2dup |  | Splice site | 1 | (Bindu *et al.*, 2011) |