Title: Dent Disease *GeneReview* – Tables 4 and 5

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Updated: September 2014

Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Table 4. Common Dent Type 1 and 2 Mutations

Gene	cDNA Change	Amino Acid Change	Mutation Type	Clinical Significance [1]	# of Families	References
CLCN5	c.100C>T	p.R34Ter	Nonsense	DP	6	Hoopes et al [1998], Cox et al [1999], Ludwig et al [2006], Tosetto et al [2006], Sekine et al [2014]
CLCN5	c.731C>T	p.S244L	Missense	HLP	10	Lloyd et al [1996], Hoopes et al [1998], Hoopes et al [2004], Tosetto et al [2006], Wu et al [2009], Sekine et al [2014]
CLCN5	c.836G>A	p.W279Ter	Nonsense	DP	4	Lloyd et al [1996], Akuta et al [1997], Lloyd et al [1997], Sekine et al [2014]
CLCN5	c.1039C>T	p.R347Ter	Nonsense	DP	7	Akuta et al [1997], Hoopes et al [2004], Ludwig et al [2005], Tosetto et al [2006], Wu et al [2009], Sekine et al [2014]
CLCN5	c.1399C>T	p.R467Ter	Nonsense	DP	5	Ludwig et al [2005], Tosetto et al [2006], Ramos-Trujillo et al [2007], Dinour et al [2009], Li & Huang [2009]
CLCN5	c.1546C>T	p.R516W	Missense	HLP	8	Akuta et al [1997], Hoopes et al [2004], Ludwig et al [2005], Wu et al [2009], Sekine et al [2014]

Gene	cDNA Change	Amino Acid Change	Mutation Type	Clinical Significance [1]	# of Families	References
CLCN5	c.1909C>T	p.R637Ter	Nonsense	DP	12	Takemura et al [2001], Cheong et al [2005], Ludwig et al [2005], Tosetto et al [2006], Cho et al [2008], Dinour et al [2009], Li & Huang [2009], Sekine et al [2014]
CLCN5	c.1942C>T	p.R648Ter	Nonsense	DP	6	Lloyd et al [1996], Lloyd et al [1997], Igarashi et al [1998], Cox et al [1999], Tosetto et al [2006], Frishberg et al [2009]
CLCN5	c.2152C>T	p.R718Ter	Nonsense	DP	5	Carballo-Trujillo et al [2003], Hoopes et al [2004], Grand et al [2009], Wu et al [2009]
OCRL	c.952C>T	p.R318C	Missense	HLP	7	Hoopes et al [2005], Utsch et al [2006], Sekine et al [2007], Böckenhauer et al [2012], Hichri et al [2011], Sekine et al [2014]
OCRL	c.1477C>T	p.R493W	Missense	HLP	4	Sekine et al [2007], Bockenhauer et al [2012], Sekine et al [2014]

1. DP: definitely pathogenic, HLP: highly likely pathogenic based on *in silico* scoring (AlignGVGD, PolyPhen-2, SIFT)

Table 5. Multiexonic/Whole-Gene Deletions for Dent Disease

Description	Phenotype	Reference
***CLCN5* gross deletions**		
2 kb (described at genomic DNA level)	Nephrolithiasis, hypercalciuric	Unwin et al [1996]
23 bp nt. 2183 (described at cDNA level)	Dent disease	Yamamoto et al [2000]
<515 kb entire gene (described at genomic DNA level)	Dent disease	Fisher et al [1994]
>180 kb incl. entire gene (described at genomic DNA level)	Dent (Japan) disease	Akuta et al [1997]
incl. entire gene (described at genomic DNA level)	Dent disease	Santo et al [2004]
incl. ex. 4-6 (described at cDNA level)	Dent disease	Igarashi et al [2000]
incl. ex. 5-8 (described at genomic DNA level)	Rickets, hypophosphataemic	Morimoto et al [1998]
OCRL		
1.2 kb incl. ex. 14 (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	Lin et al [1997]
E4I4+3 kb to E8I8+857 (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	Monnier et al [2000]
E7I7+135 to E12I12+347 (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	Monnier et al [2000]
Entire gene (described at cDNA level)	Lowe oculocerebrorenal syndrome	Peverall et al [2000]
ex. 20-24 (described at genomic DNA level)	Dent disease	Hichri et al [2011]
ex. 3-4 (described at genomic DNA level)	Dent disease	Hichri et al [2011]
ex. 6-12 (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	LOVD Database [2005]
incl. ex. 1-3 (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	Monnier et al [2000]
incl. ex. 1-4 (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	Monnier et al [2000]
~4 Mb incl. entire gene (described at genomic DNA level)	Lowe oculocerebrorenal syndrome	Addis et al [2007]

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