HOKKAIDO UNIVERSITY

Title	Onset mechanism of a female patient with Dent disease 2
Author(s)	Okamoto, Takayuki; Sakakibara, Nana; Nozu, Kandai et al.
Citation	Clinical and experimental nephrology, 24, 946-954 https://doi.org/10.1007/s10157-020-01926-4
Issue Date	2020-10
Doc URL	https://hdl.handle.net/2115/82888
Rights	This is a post-peer-review, pre-copyedit version of an article published in Clinical and Experimental Nephrology. The final authenticated version is available online at: http://dx.doi.org/10.1007/s10157-020-01926-4
Type	journal article
File Information	Supplementary figure 2rr.pdf

Hokkaido University Collection of Scholarly and Academic Papers : HUSCAP

Suplementary figure 2

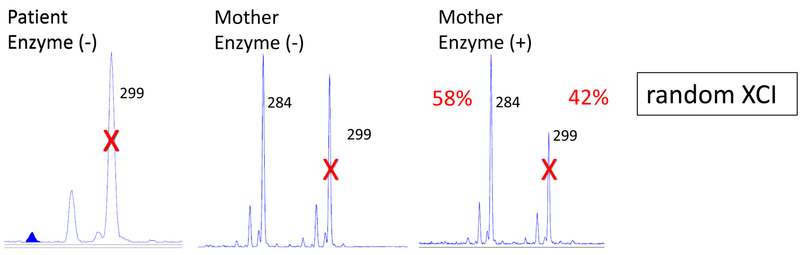
a

Pt	Age	Mutation	Location	Consequence	Cataract	Intellectual disability	Ambulation	Renal tubular acidosis	Cr-eGFR
D93	0	* Exon12-15deletion	-	large deletion	+	Severe	-	+	131.44
D100	4	* c.1495G>A	Exon15	p.(Asp499Asn)	+	Mild	+	-	77.69
D113	15	* c.454G>T	Exon7	p.(Glu152*)	+	Border line	+	-	93.70

b

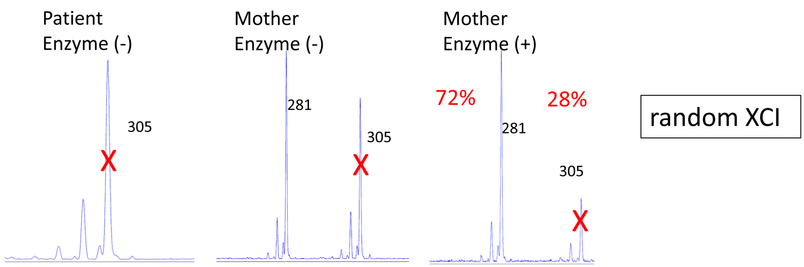
D93 : severe (typical) phenotype

Pt	Age	Mutation	Location	Consequence	Cataract	Intellectual disability	Ambulation	Renal tubular acidosis	Cr-eGFR
D93	0	* Exon12-15deletion	-	large deletion	+	Severe	-	+	131.44



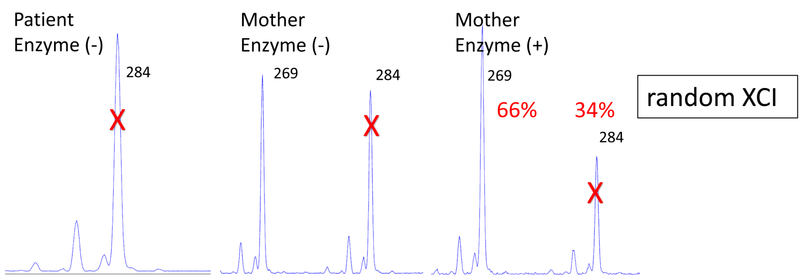
D100 : mild phenotype

Pt	Age	Mutation	Location	Consequence	Cataract	Intellectual disability	Ambulation	Renal tubular acidosis	Cr-eGFR
D100	4	* c.1495G>A	Exon15	p.(Asp499Asn)	+	Mild	+	-	77.69



D113 : mild phenotype

Pt	Age	Mutation	Location	Consequence	Cataract	Intellectual disability	Ambulation	Renal tubular acidosis	Cr-eGFR
D113	15	* c.454G>T	Exon7	p.(Glu152*)	+	Border line	+	-	93.70



c

Summary of XCI

Patient	XCI ratio	XCI
D93 mother	42:58	Random XCI
D100 mother	28:72	Random XCI
D113 mother	34:66	Random XCI
our patient (blood)	85:15	Skewed XCI
our patient (urine)	81:19	Skewed XCI

Mutation allele