

# ERRATUM

In the February 2005 issue of the *Journal*, in the article entitled “Dent Disease with Mutations in *OCRL1*” by Hoopes et al. (76:260–267), table 1 contains two errors in the “Nucleotide Change” column. For family 24, the correct nucleotide change for the mutation is 901C→T. For family 26, the correct nucleotide change for the mutation is 510-2A→G. We have also modified the notation in the “Nucleotide Change” column for families 25 and 29, to follow the guidelines for mutation nomenclature found at the

Human Genome Variation Society Web site (<http://www.genomic.unimelb.edu.au/mdi/mutnomen/>) and in the work of den Dunnen and Antonarakis.<sup>1</sup> The authors regret the errors.

## Reference

- den Dunnen JT, Antonarakis SE (2000) Mutation nomenclature extensions and suggestions to describe complex mutations: a discussion. *Human Mutation* 15:7–12

**Table 1. Mutations and Expression Data in Affected Males from the Five Families**

Family	Patient's Age at Examination (years)	PIP <sub>2</sub> Phosphatase Activity <sup>a</sup> (nmol/min/mg)	Level of OCRL Protein (Western Blot) <sup>b</sup>	Exon	Mutation Type	Nucleotide Change	Effect on Translation	Screening Method	No. of Normal X Chromosomes Screened
20	9	.63	++	14	Substitution	c.1385A→G	Y462C (Tyr→Cys)	RFLP ( <i>Tsp451</i> )	120
24 <sup>c</sup>	22/27	.76/.85	+++ / ++++	11	Substitution	c.901C→T	R301C (Arg→Cys)	RFLP ( <i>Hha1</i> )	132
25	8	.42	–	7	2-base insertion	c.438_439dupAA	I147K, Stop	Allele-specific PCR	106
29	10	.49	–	5	4-base deletion	c.261_264delTTTG	C87Stop	RFLP ( <i>Eco571</i> )	106
26	9	.54	–	Intron 7 (5' of exon 8)	Splice-site mutation	c.510-2A→G	Unknown (likely frameshift)	RFLP ( <i>BfaI</i> )	112

<sup>a</sup> Control fibroblast, 4.71 nmol/min/mg; Lowe fibroblast, 0.52 nmol/min/mg.

<sup>b</sup> Control fibroblast, ++++; Lowe fibroblast, –.

<sup>c</sup> There were two affected brothers in this family.