Supplementary Table S6: Subset of Reported Human *CRX* Mutations Demonstrating the Heterogeneous Nature of the Disease Genotype and Phenotype.

Phenotype	Nucleotide Mutation	Mode of Inheritance	Amino Acid Mutation	Missense Location	Effect of PTC	References
LCA	24-25 ins G	recessive	P9A FS +60 and PTC		Suspected Null Allele	77, 78
CORD	C121T	dominant	R41W	homeodomain		58
RP	G122A	dominant	R41Q	homeodomain		51, 57, 79
CORD*	G122A	dominant	R41Q	homeodomain		58
CORD	G238A	dominant	E80K	homeodomain		80
CORD	A239C	dominant	E80A	homeodomain		51, 53
LCA	G268A	recessive	R90W	homeodomain		55
RP	G324A	dominant	R115Q	Basic domain		81
LCA	T438-T449 del 12	dominant	P147-S150 deletion			51,81, 82
CORD	G502 del 1	dominant	E168S FS +17 and PTC	WSP	eliminates TTD 1 & 2	53, 83
LCA	G502-503 del 2	dominant	E168V FS +3 and PTC	WSP	eliminates TTD 1 & 2	83, 84
LCA	T510 del 1	dominant	P170P FS +15 and PTC	WSP	eliminates TTD 1 & 2	85
LCA	T571del1	dominant	Y191M FS +1 and PTC		eliminates TTD 1 & 2	57
CORD	C585 ins C	dominant	A196R FS +38 and PTC		eliminates TTD 1 & 2	51, 79
CORD	C587-C590 del 4	dominant	A196G FS +20 and PTC		eliminates TTD 1 & 2	58
CORD	G724A	dominant	V242M	TTD 1		58

PTC premature termination codon; FS frame shift; TTD translational transactivation domain. TTD locations based on Chen et al, (41).
*Later studies by Rivolta, et al., (57) suggested that the phenotype should be identified as RP.