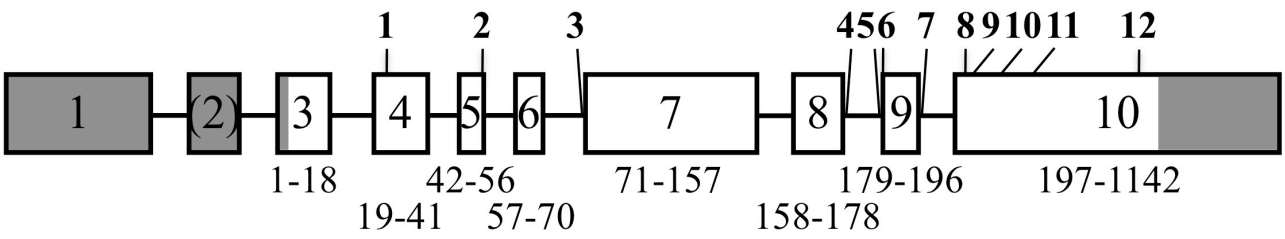
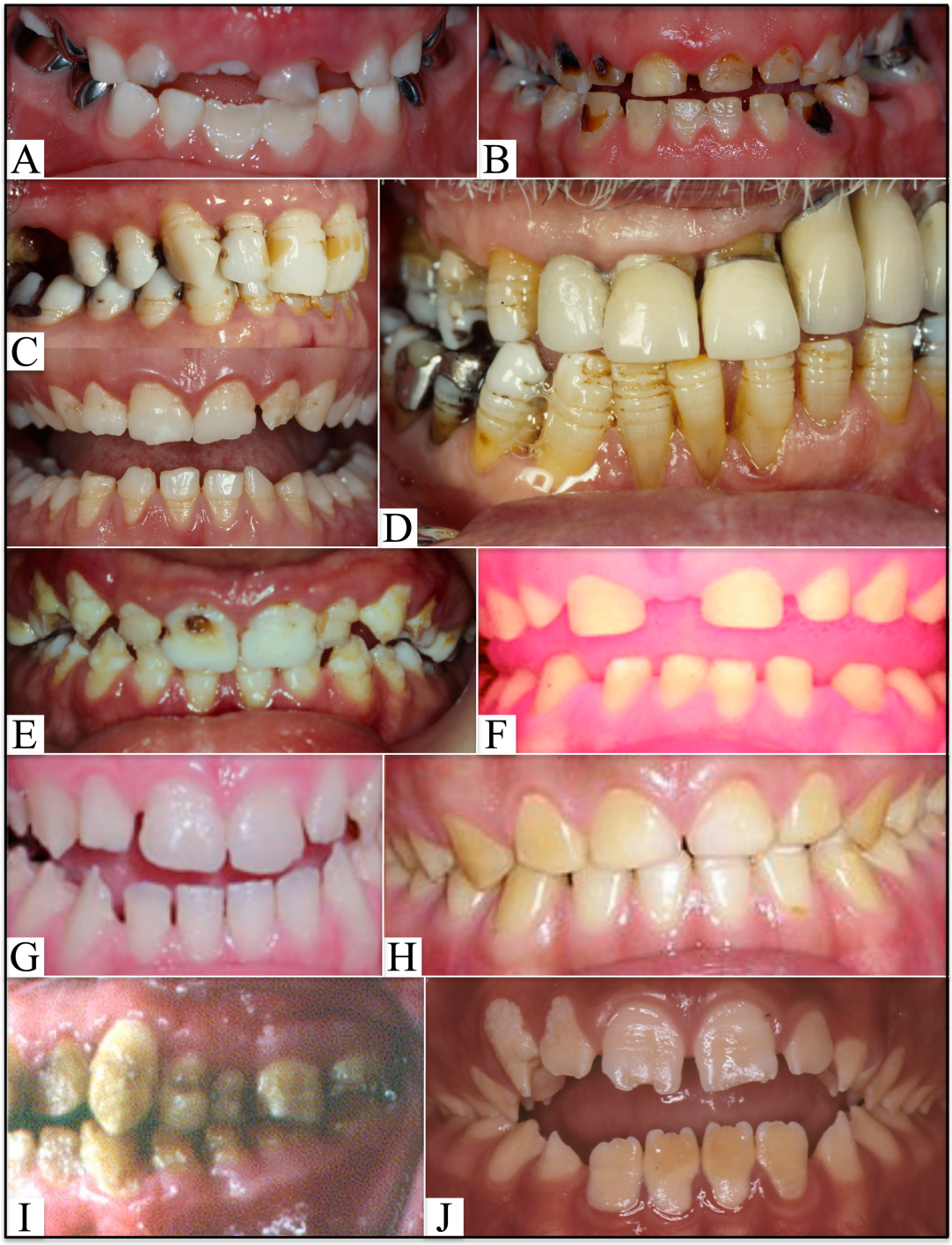


#	Gene	cDNA	Protein	References
1	g.2979delA	c.107delA	p.N36Ifs56	Here
2	g.3139A>T	c.157A>T	p.K53X	(1, 2)
3	g.5563A>C	c.211-2A>C	p.M71_Q157del	(3)
4	g.7152G>A	c.534+1G>A	p.A158_Q178del	(4)
5	g.9045A>G	c.535-2A>G	p.R179-N196del	(5, 6)
6	g.9048G>T	c.536G>T	p.R179M	(7)
7	g.(9095-9101)delG	c.(583-588+1)delG	p.R179-N196del	(3, 8-10)
8	g.13330 C>T	c.647G>T	p.S216L	(11)
9	g.13420C>A	c.737C>A	p.S246X	(12)
10	g.13703_13704insAGTCAGTACCAGTACTGTGTC	c.1020_1021insAGTCAGTACCAGTACTGTGTC	p.V340_M341insSQYQYCV	(12)
11	g.13942_13943/insAG	c.1259_1230insAG	p.V422PfsX448	(10-15)
12	g.15674delT	c.2991delT	p.L998WfsX1062	(13)

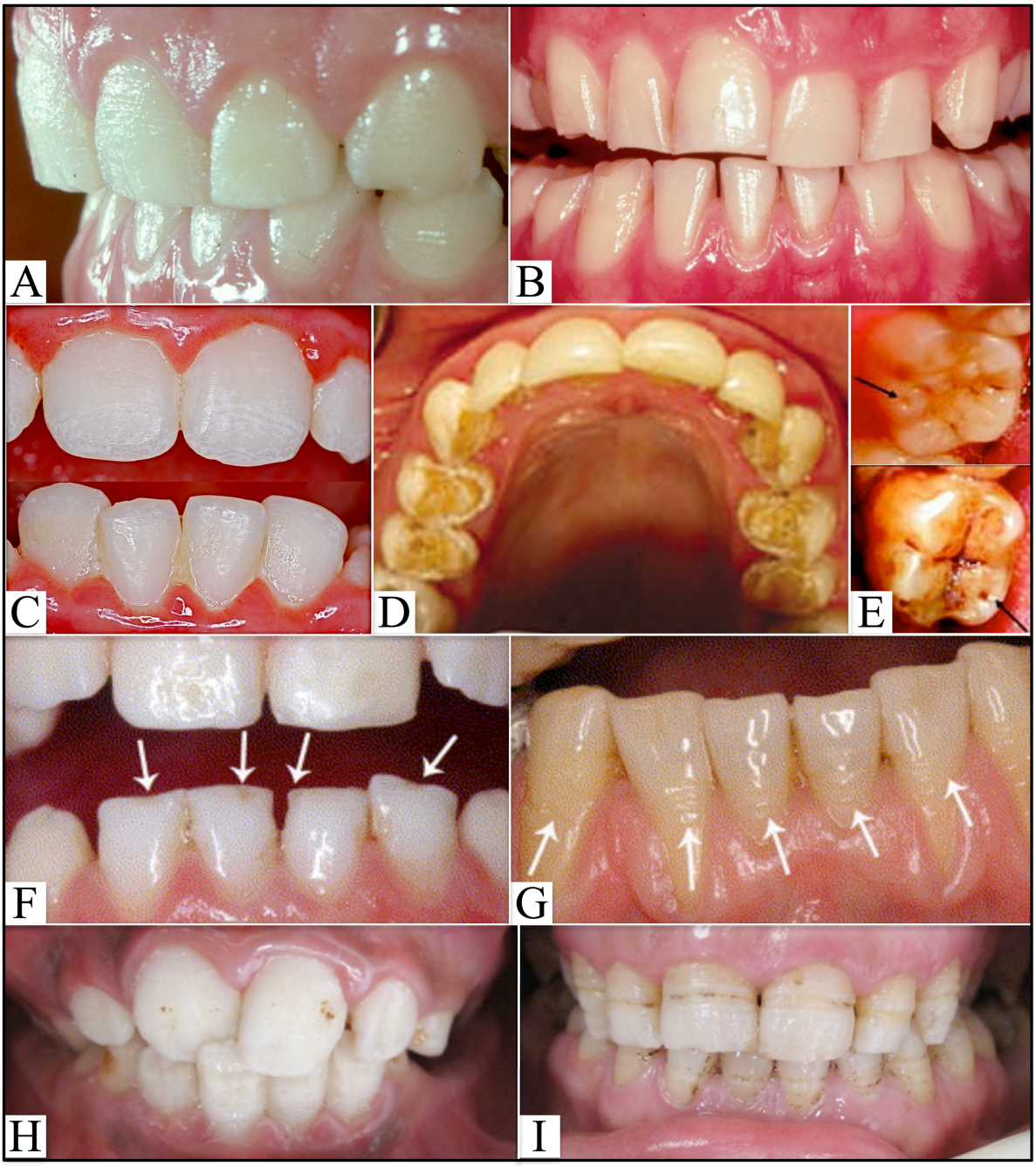
Both *ENAM* Alleles Affected

#	Allele 1	Allele 2	References
1	p.S216L	p.V422PfsX448	(11)
2	p.V340_M341insSQYQYCV	p.V422PfsX448	(12)
3	p.V422PfsX448	p.V422PfsX448	(13-15)

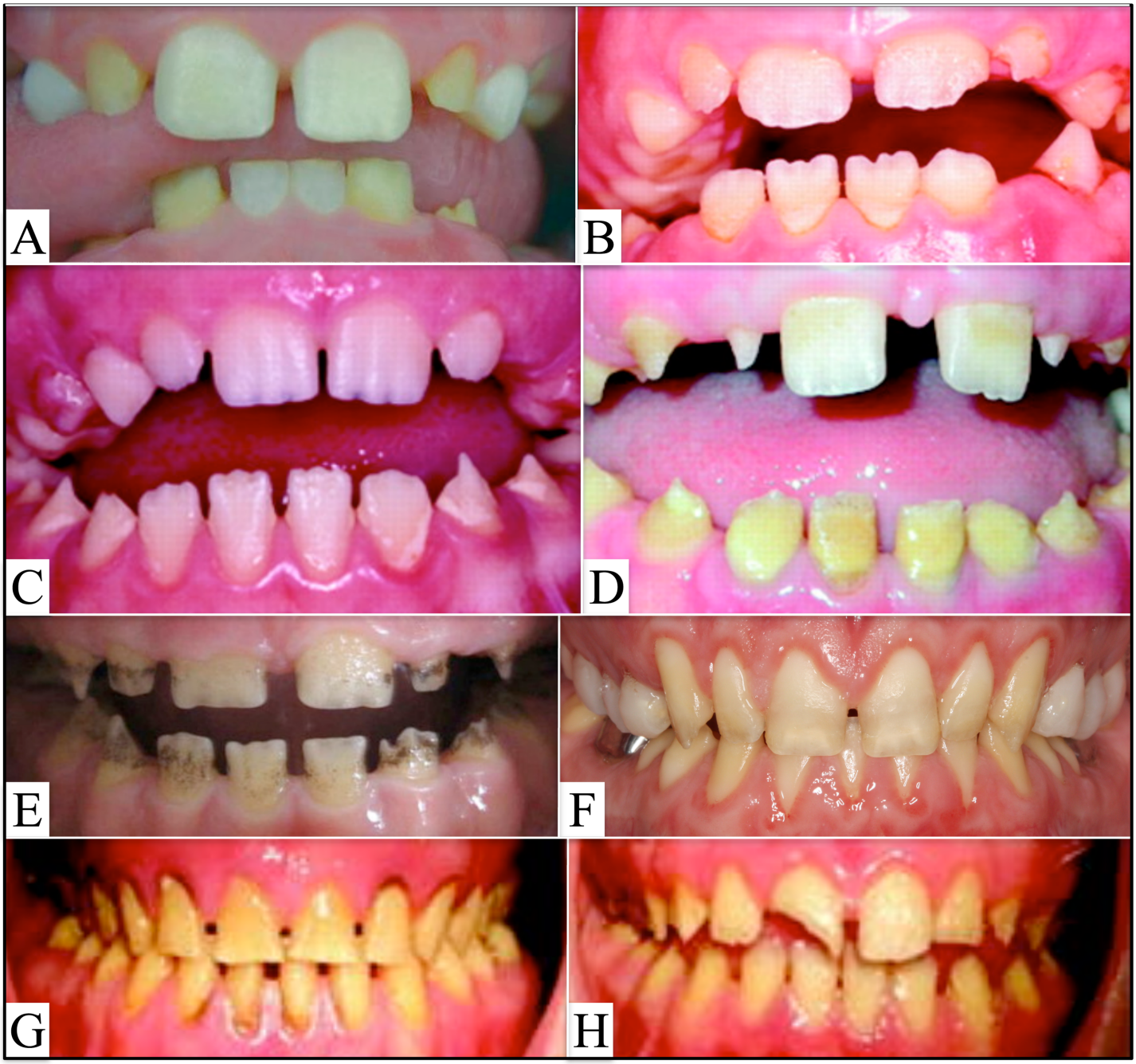
Supplemental Figure 1. *ENAM* gene structure and disease-causing mutations. The numbered boxes are exons; the lines introns. The numbers below each exon show the range of amino acids encoded by it. Shaded exon regions are non-coding. Bold numbers indicate *ENAM* mutations. The gene numbers start from the first nucleotide of the *ENAM* reference sequence NG_013024.1. The cDNA numbers start from the translation initiation site of *ENAM* reference sequence NM_031889.2. The protein designations are deduced and have not been verified experimentally.



Supplemental Figure 2. Oral photos of phenotypes associated with defined heterozygous *ENAM* defects. ***A-B:*** c.107delA exon 4 frameshift (this report), ***C-D:*** c.157A>T exon 5 nonsense mutation (1) with permission from Oxford University Press; (2) with permission from John Wiley & Sons A/S, ***E:*** c.211-2A>C intron 6 splice junction acceptor (3), ***F:*** c.534+1G>A intron 8 splice junction donor (4) with permission of Oxford University Press, ***G-H:*** c.535-2A>G intron 8 splice junction acceptor (5) with permission from Kager Publishers (6), ***I:*** c.536G>T first codon in exon 9 missense mutation (p.R179M), (7) with permission of Elsevier, ***J:*** c.(583-588+1)delG exon 9 frameshift/intron 9 splice junction donor (3).



Supplemental Figure 3. Oral photos of phenotypes associated with defined 5' *ENAM* defects. ***A-B:*** c.(583-588+1)delG exon 9 frameshift/intron 9 splice junction donor (9) with permission from Elsevier, ***C:*** c.647G>T exon 10 phosphoserine missense mutation (p.S216L) (11), ***D:*** c.737C>A exon 10 nonsense mutation (p.S246X) (12) reprinted by permission of SAGE Publications, ***E:*** c.1020_1021insAGTCAGTACCAGTACTGTGTC exon 10 insertion (p.V340_M341insSQYQYCV) (12) reprinted by permission of SAGE Publications, ***F-G:*** c.1259_1230insAG exon 10 frameshift (p.V422PfsX448) (10) with permission from Elsevier, ***H-I:*** c.2991delT exon 10 frameshift (p.L998WfsX1062) (13) reprinted by permission of SAGE Publications.



Supplemental Figure 4. Oral photos of phenotypes associated with *ENAM* defects in both alleles. In all cases reported so far, one *ENAM* allele has always been the p.V422PfsX448 exon 10 frameshift. ***A-E:*** exon 10 frameshift (p.V422PfsX448) in both alleles (13) reprinted by permission of SAGE Publications; (14) with permission from BMJ Publishing Group Ltd; (15) with permission of the American Academy of Pediatric Dentistry, ***F:*** p.S216L/p.V422PfsX448 compound heterozygote (11), ***G-H:*** p.V340_M341insSQYQYCV/p.V422PfsX448 compound heterozygote (12) reprinted by permission of SAGE Publications. In all cases of homozygous *ENAM* defects the phenotype is severe, generalized enamel hypoplasia.

Supplemental Data References

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