A Novel *AMELX* Mutation, Its Phenotypic Features, and Skewed X Inactivation

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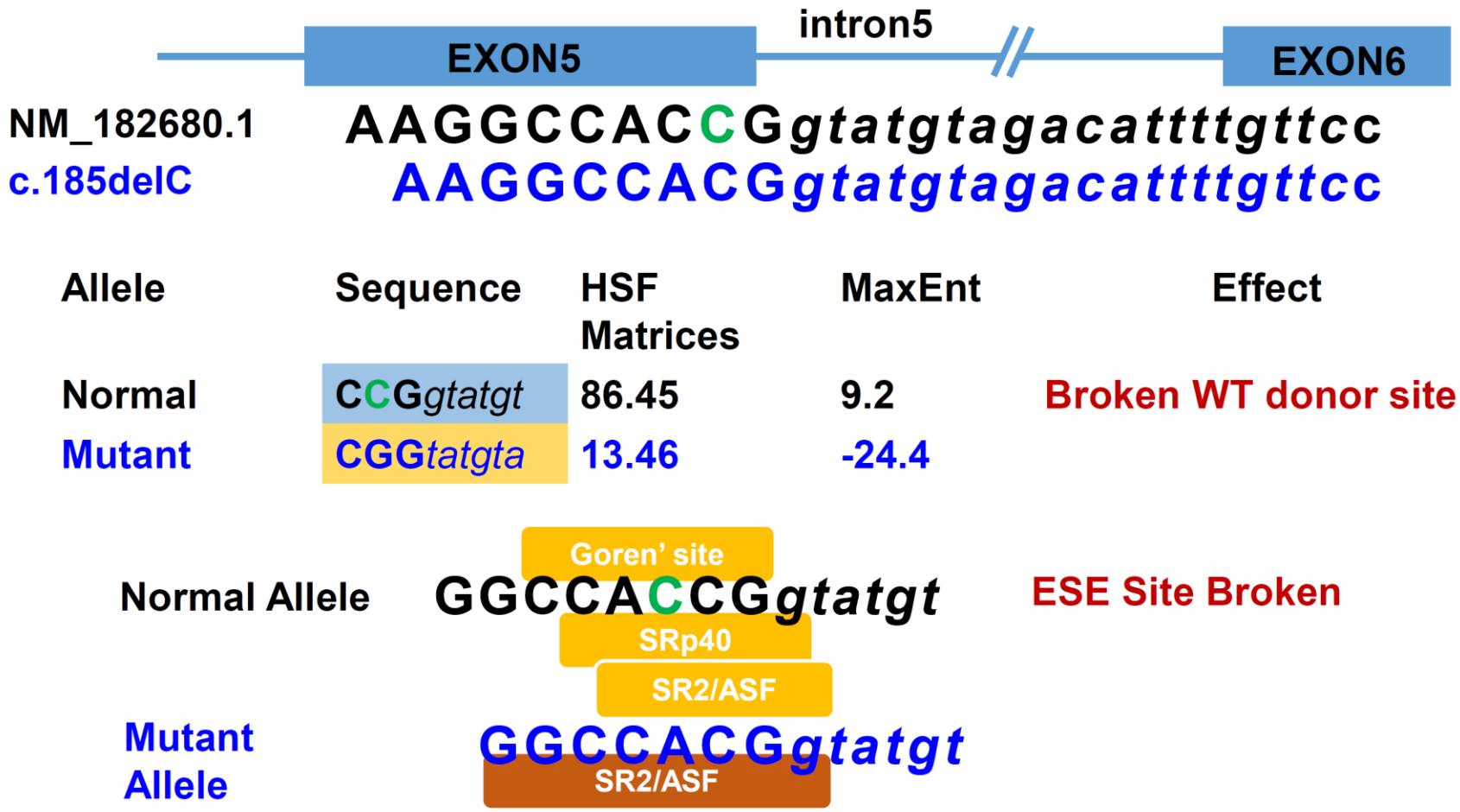
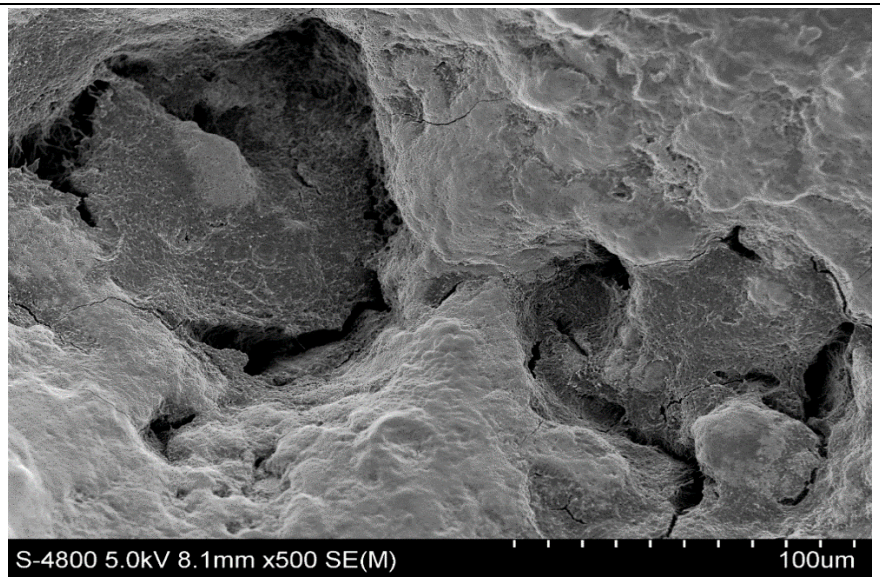
Appendix



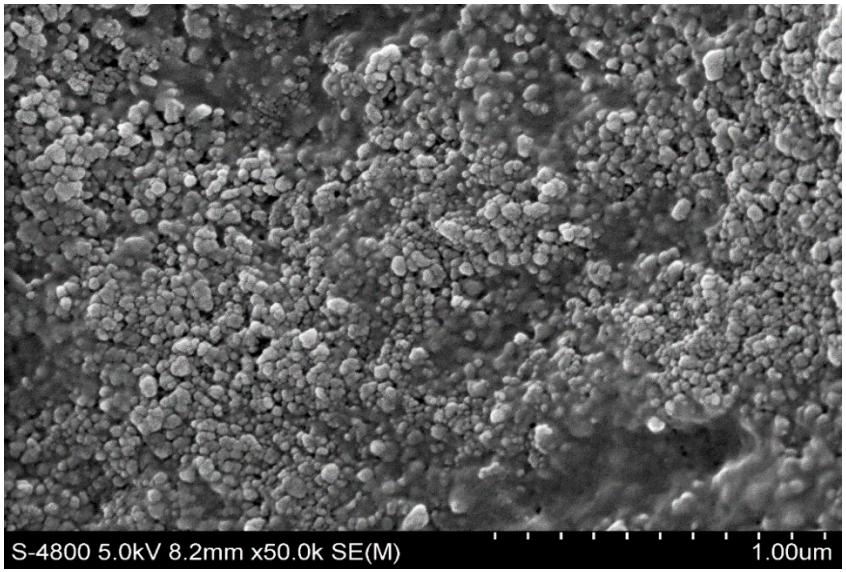
Appendix Figure 1. Splicing analysis of c.185delC

The deletion of C at c.185 is predicted to destroy the wild type (WT) donor site and produce new donor site. Splicing analysis. HSF: Human Splicing Finder; HSF Matrices represent Position Weight Matrices; MaxEnt: Maximum Entropy; ESE: Exonic Splicing Enhancers. SRp40, SR2/ASF are splicing factors.

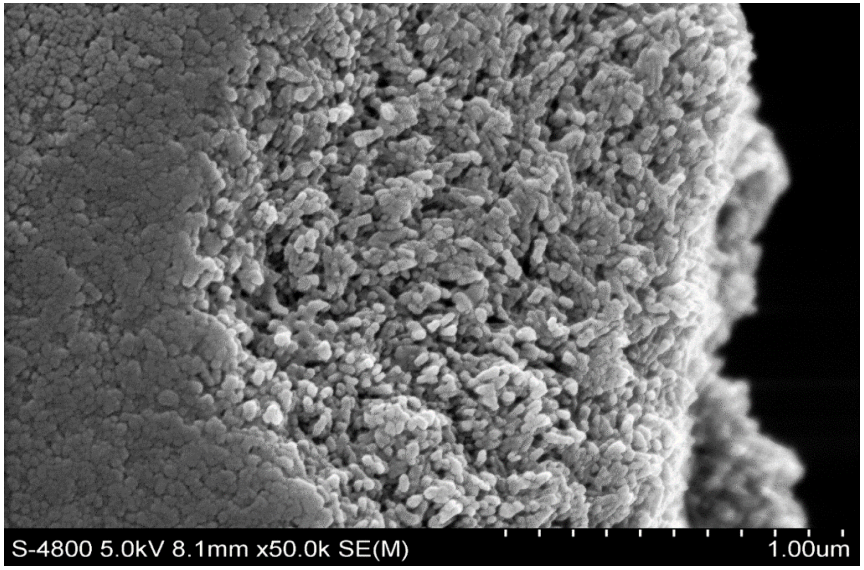
c


S-4800 5.0kV 8.1mm x500 SE(M)
100um

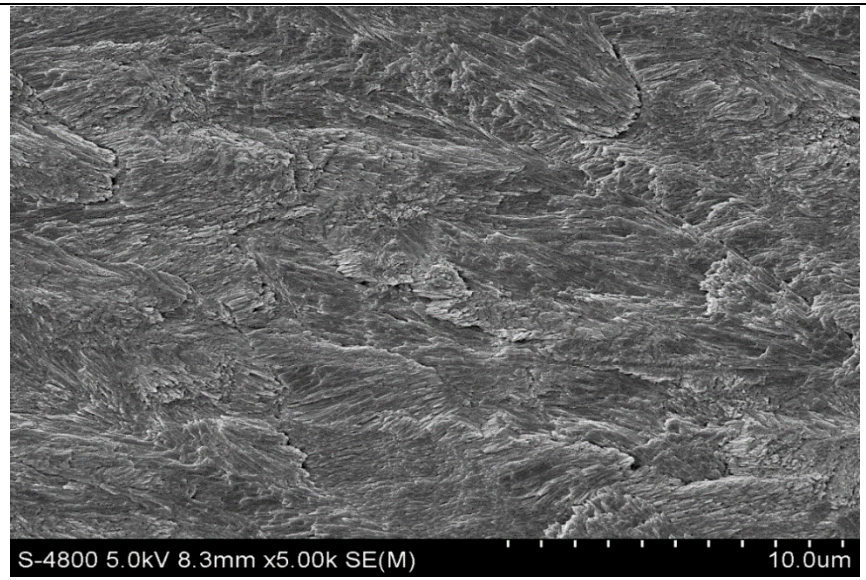
d 2

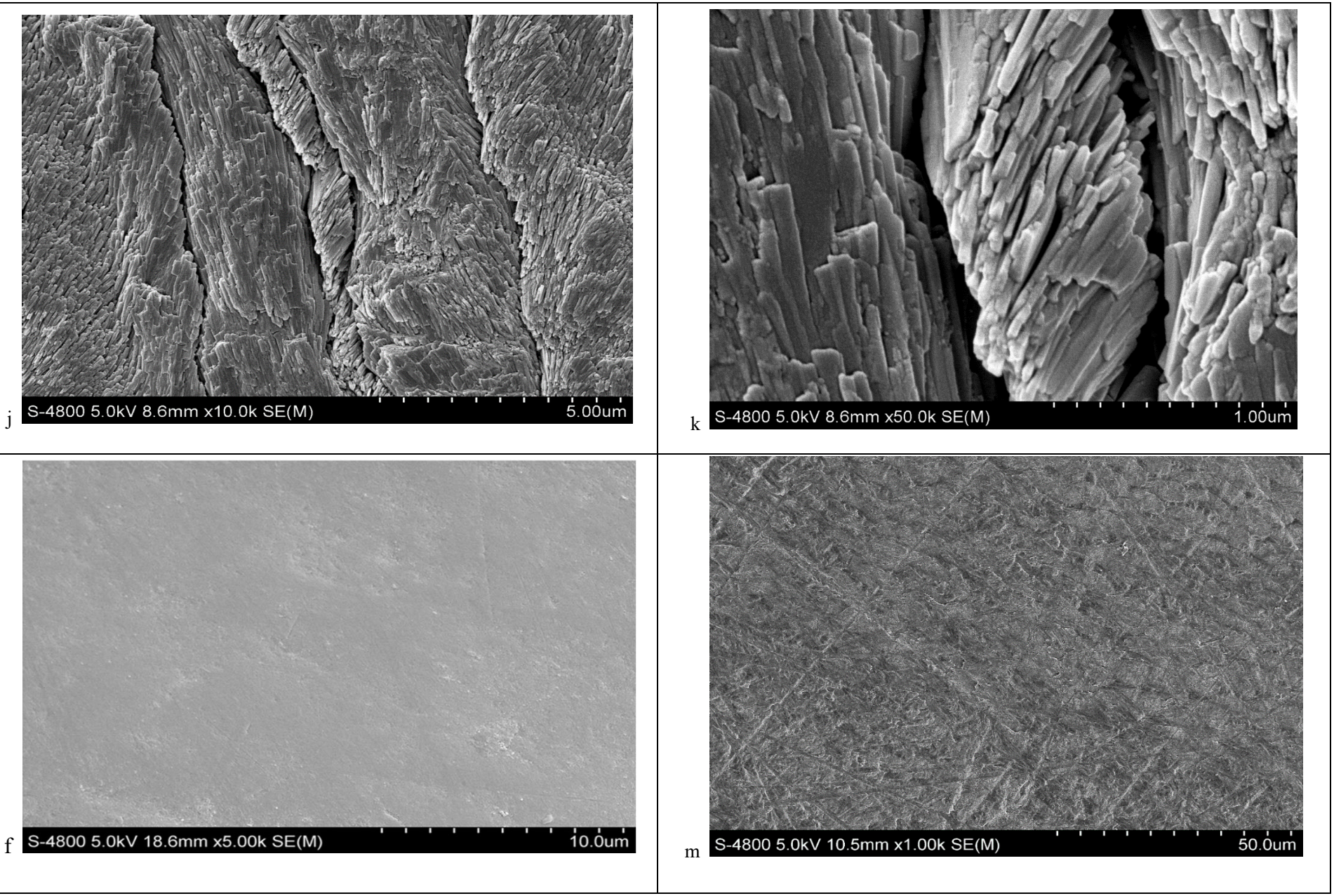

S-4800 5.0kV 8.2mm x50.0k SE(M)
1.00um

e


S-4800 5.0kV 8.1mm x50.0k SE(M)
1.00um

i


S-4800 5.0kV 8.3mm x5.00k SE(M)
10.0um



Appendix Figure 2. High resolution SEM images of the third molar of the proband's mother and the control in figure 4.

c. Original SEM image of figure 4c showing the large pits on the surface of enamel.

d2. Original SEM image of figure 4d2 showing the rough area of small pits on the surface of enamel.

e. Original SEM image of figure 4e showing the exposed crack region of enamel.

i, j, k. Original SEM images displaying the densely packed enamel prism region (i) and the rough, uneven and exposed enamel prisms with some cracks (j). Panel k is a part of magnification of image j.

f. Original SEM image of figure 4f showing the control enamel surface.

m. Original SEM image showing the cross section of control enamel, not shown in figure 4.