Introduction: Biomarkers that identify individuals at an early stage of Alzheimer's disease (AD) would be useful as this would allow timely preventative intervention. There is increasing interest in the evaluation of chromosome damage markers within somatic cells of neurodegeneration patients.

Aims: Whether micronuclei and other parameters of genome damage and cell death in the buccal mucosa could be used as a non-invasive method of identifying individuals at risk of developing AD.

Method: A cytome approach was adopted in which various cell types and their ratios were quantified to identify those parameters most strongly associated with AD. Buccal cells were classified into categories that distinguish between “normal” cells and cells considered “abnormal”, based on nuclear morphology. Abnormal nuclear morphologies are thought to be indicative of DNA damage or cell death.

Results: Frequencies of basal cells (p< 0.0001), condensed chromatin cells (p< 0.0001) and karyorrhectic cells (p< 0.0001) were found to be significantly lower in Alzheimer's patients. These changes may reflect alterations in the cellular kinetics or structural profile of the buccal mucosa, and may be useful as potential biomarkers in identifying individuals with a high risk of developing AD. The odd's ratio of being diagnosed with AD for individuals with a basal plus karyorrhectic cell frequency < 41 per 1000 cells is 140, with a specificity of 97% and sensitivity of 82%.

Conclusions: These promising results need to be replicated in cohorts of other neurodegenerative disorders to determine specificity of changes to Alzheimer's patients.