HLA-G 14bp Polymorphism in Autism

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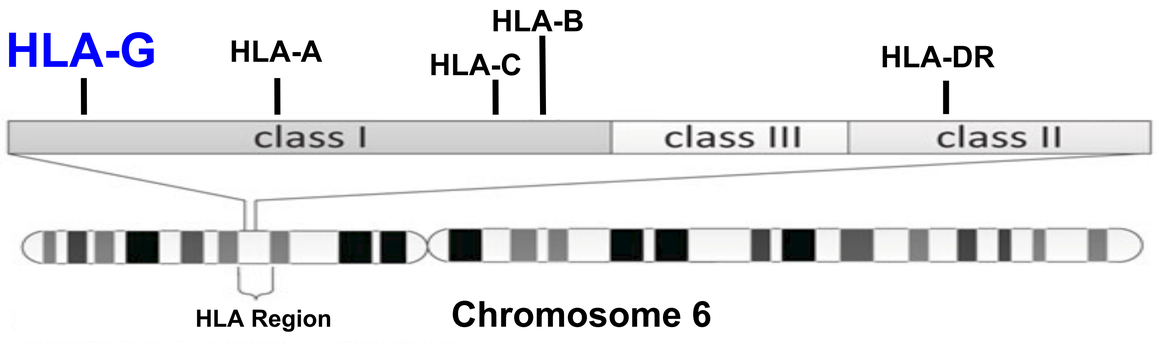
Introduction

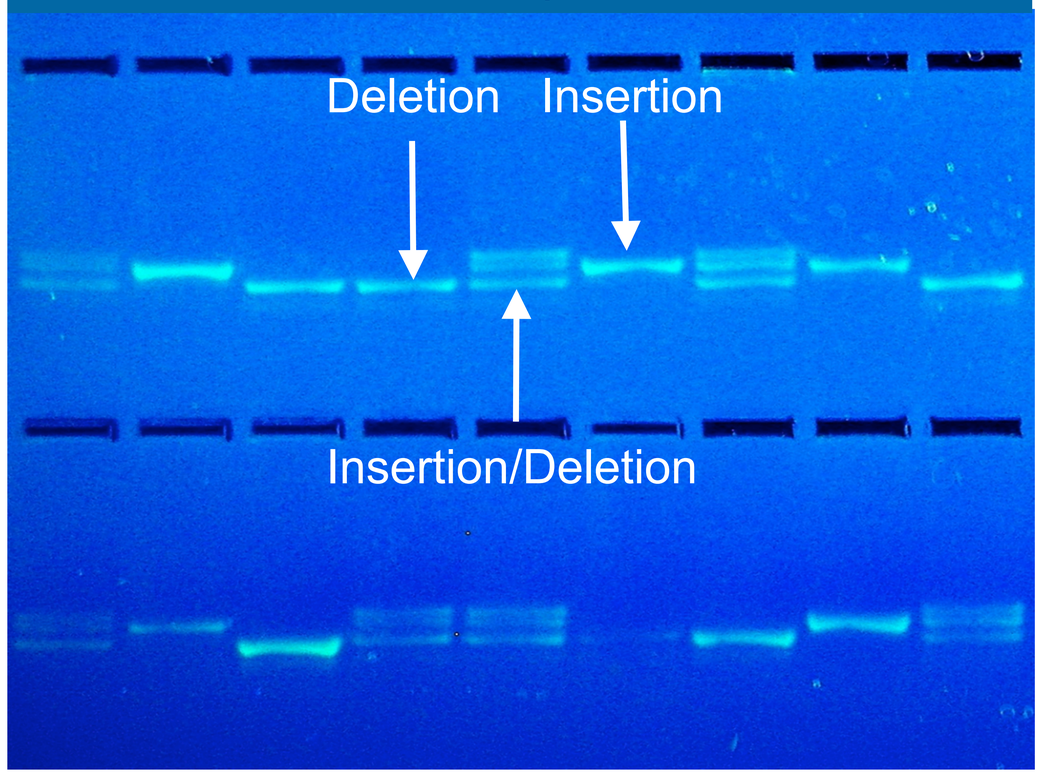
Autism Spectrum Disorder (ASD) is a neurodevelopment disorder characterized by deficits in communicative and social behaviors (Meltzer, 2017) . As of 2012 the CDC reported that 1 of 68 children born in the U.S. have ASD (Christensen, 2016).

The immune systems of mother and child can be important in ASD. A signaling molecule, HLA-G, helps regulate maternal natural killer cell interaction with the fetus. A defect in HLA-G could increase NK cell activity, leading to abnormal neurodevelopment in the fetus (Carosella, 2008).

Our study focuses on a 14 base pair insertion/deletion found in the HLA-G gene of autistic subjects and their mothers, previously examined in an Italian population by Guerini (2014). We are also expanding to look at HLA-G and intellectual disability (ID) in ASD. HLA-DRB1, another gene in the HLA region of chromosome 6, has been linked to ASD and impaired ID (IQ<80, Wang, 2013).



Gel Electrophoresis



Methods

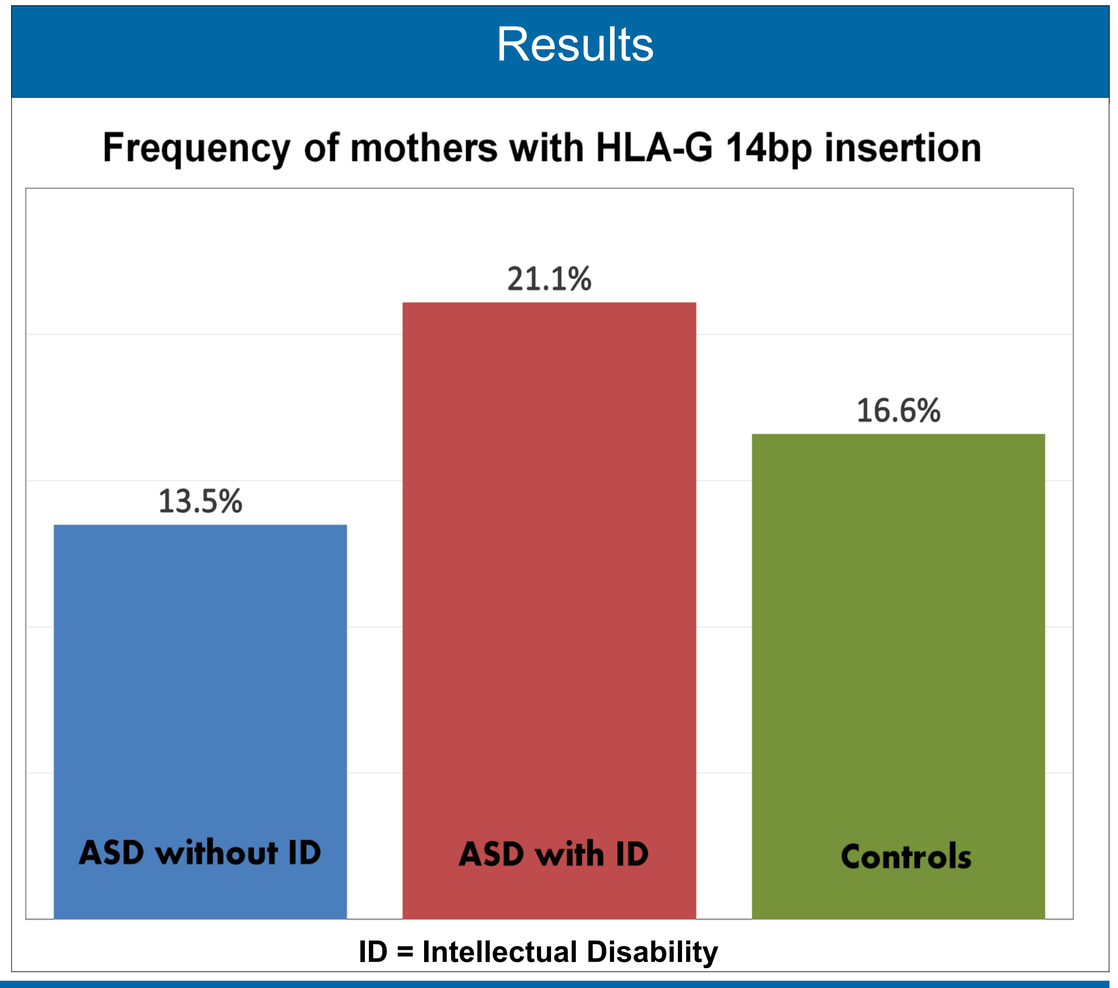
DNA from 259 subjects of the Early Markers for Autism (EMA) projects was genotyped for the HLA-G 14bp insertion/deletion.

Mothers of ASD with ID = 38
Mothers of ASD without ID = 52
Mothers of control subjects = 169

Genotyping was done by PCR and gel electrophoresis. A 14 base pair difference in PCR product size indicates a deletion or insertion in the DNA sequence

Autistic children are still in the process of being genotyped.

Results



Conclusion

HLA-G 14bp insertion increased in mothers of ASD with ID compared to controls and ASD without ID.

Although results for HLA-G genotyping in the mothers has not proven to be statistically significant, our population size is small.

Larger scale studies may be needed to show the significance of HLA-G polymorphisms and intellectual disability in autism.

We are still awaiting results for HLA-G genotyping in the autistic children.

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