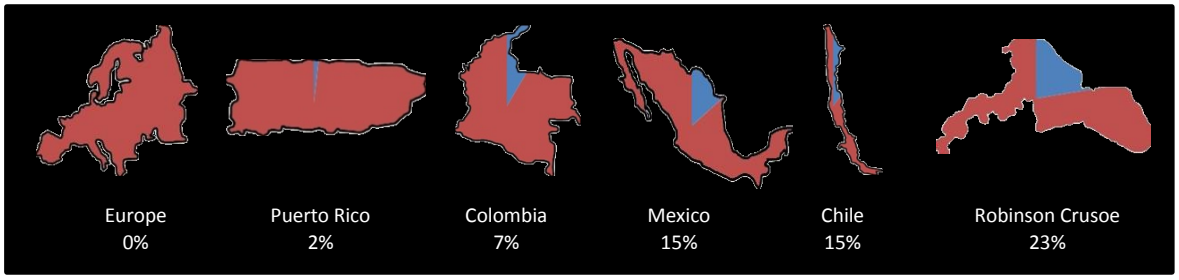
Investigation of the Robinson Crusoe Population

In this study, we looked at genetic variation in a particular population of people affected by language impairment. This population live on the Robinson Crusoe Island in Chile.

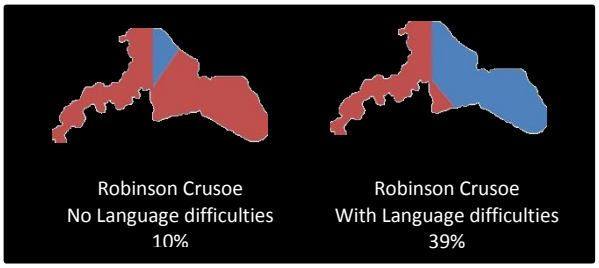
The Robinson Crusoe Island was first inhabited by a group of 64 Founders in 1876. Because the Island is hard to get to, most of the people who live there today are related to these Founders making the Island like one big family.

We are interested in the population because lots of the Islanders have problems with speech and language development. We think this might be because some of the Founders had mutations which made it harder to learn to use language. These mutations would have been inherited by most of the Islanders explaining why such a high number of the current population have speech and language difficulties.

In order to test this hypothesis, we looked at the DNA sequence of all known genes in 5 Islanders affected by speech and language problems. We looked for changes in the genetic sequence that were shared by all 5 individuals. Then we looked at the rest of the population to see whether any of these changes were correlated to the presence of language impairment. We found one genetic change that was more common in Islanders affected by language disorders than in Islanders with typical language development.



In this study, we found a change in the genetic sequence that was particularly common in people living on the Robinson Crusoe Island. The bluer the country, the more common the variant. The numbers under the country show the percentage of the population estimated to carry the variant.



More importantly, we found that this variant was particularly common in inhabitants of the Robinson Crusoe Island who were affected by language impairment.

When we looked at this gene in people from the UK, we again found that mutations in the gene were more common in individuals affected by language impairment than those with typical language development.

The identified gene is called *NFXL1* and it is responsible for turning other genes on and off within cells. We hope that further investigation of this gene and the genes that it controls, will provide a better understanding of the biological foundations of language development.

This summary refers to the following research paper: Genome-wide analysis identifies a role for common copy number variants in specific language impairment. Exome Sequencing in an Admixed Isolated Population Indicates NFXL1 Variants Confer a Risk for Specific Language Impairment. Villanueva P, Nudel R, Hoischen A, Fernández MA, Simpson NH, Gilissen C, Reader RH, Jara L, Echeverry MM, Francks C, Baird G, Conti-Ramsden G, O'Hare A, Bolton PF, Hennessy ER, the SLI Consortium, Palomino H, Carvajal-Carmona L, Veltman JA, Cazier JB, De Barbieri Z, Fisher SE, Newbury DF. PLoS Genetics. 2015 March 17 [Epub ahead of print] PMID:25781923 [pdf]