Abstract: Deciphering the genetics of speech and language disorders

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Innovations in molecular biology have raised the possibility of identifying genetic risk variants that confer susceptibility to developmental language disorders. In 2001, my colleagues and I discovered an intriguing gene known as *FOXP2*. Children with mutations of *FOXP2* have problems mastering sequences of co-ordinated mouth movements needed for fluent speech, accompanied by expressive and receptive impairments of spoken and written language. *FOXP2* disruption explains only a small percentage of cases of speech disorder. Nevertheless, studies of this gene give a novel route for determining the relevant neuronal mechanisms. *FOXP2* encodes a regulatory protein which switches on and off other genes. It is evolutionarily ancient, present in similar form in diverse vertebrate species, where it helps regulate development and function of certain circuits in the brain. Analyses of the gene in primates indicate that it underwent accelerated change on the human lineage after splitting from the chimpanzee. However, *FOXP2* should not be viewed as the mythical “gene for speech”, but instead as one piece of a complex puzzle.

My talk will describe how *FOXP2* is being used as a unique window into key neurogenetic pathways. To this end, researchers exploit a wide range of systems, from brain cells grown in the laboratory, to animal models. For example, state-of-the-art techniques are being employed to identify genes that *FOXP2* regulates in the human brain (its downstream targets). Remarkably, some of these targets are themselves implicated in common language-related disorders. Studies of *FOXP2* in animals and birds suggest that it is important for maintaining plasticity of brain circuits. Overall, this body of work may ultimately yield improved diagnosis and treatment of speech- and language-related disorders. It is also shedding the first light on how the human genome helps to build a language-ready brain.