FOXP2 Syndrome

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Rare mutations of the FOXP2 transcription factor gene cause a monogenic syndrome characterized by impaired speech development and linguistic deficits. Recent genomic investigations indicate that its downstream neural targets make broader impacts on common language impairments, bridging clinically distinct disorders. Moreover, the striking conservation of both FoxP2 sequence and neural expression in different vertebrates facilitates the use of animal models to study ancestral pathways that have been recruited towards human speech and language. Intriguingly, reduced FoxP2 dosage yields abnormal synaptic plasticity and impaired motor-skill learning in mice, and disrupts vocal learning in songbirds. Converging data indicate that Foxp2 is important for modulating the plasticity of relevant neural circuits. This body of research represents the first functional genetic forays into neural mechanisms contributing to human spoken language.