THE GRADUATE CENTER
CITY UNIVERSITY OF NEW YORK

City University of New York

Xiaomeng Ma

A systematic review of genetic disorder and language development

INTRODUCTION

Genetic factors are important contributors to speech and language development. The specific language skill is still largely unknown, since language development is always intertwined with development of other higher cognitive functions, such as memory, executive function, etc. Meanwhile, language is a distinct cognitive domain that has its own neurological pathways, which makes it reasonable assume that there is language-specific genetic basis that does not interfere with other cognitive domains. Not all genetic language

Language Disorder ≠ Cognitive Impairment

Not all genetic language disorder causes impairment in other cognitive domains. For example, the family with mutant FOXP2 genes only have language disorders (specifically, difficulties with grammar) but no other cognitive deficits.

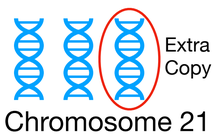
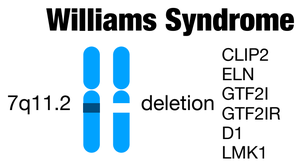
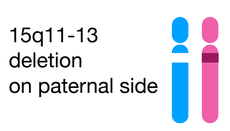
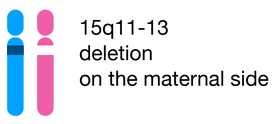
Genetic Disorder ≠ Language Impairment

Similarly, language impairment is not always in comorbid condition with other neuro-cognitive disorders a way that not all genetic disorders lead to language impairment. For example,Down Syndrome is caused by abnormality in chromosome 21 while Williams Syndrome is caused by the deletion of chromosome 7. The features of both syndrome include being hyper-social, mild intellectual disability and weak visuospatial cognitive ability. However, language impairment is only found in Down Syndrome but not in Williams Syndrome.

Purpose of this study:

This study is going to disentangle the relationship between language development and other higher cognitive functions, such as intelligence disabilities and social behaviors by reviewing cognitive profiles of nine genetic disorders (Down Syndrome, Williams Syndrome, Angolan Syndrome, Prader-Willis Syndrome, Wolf-Hirschhorn Syndrome, Smith-Magenis Syndrome, Fragile X Syndrome and Cri du chat Syndrome). Common features of these disorders include mild to moderate intellectual disabilities and difficulties in social interactions (excessive social or autistic-like). However, language and speech impairment are not found in all of these disorders. By reviewing cognitive profiles of the genetic disorders and their language development, this study hopes to explore possible directions to further investigate genetic basis for language-specific skills.

GENETIC DISORDER AND LANGUAGE DEVELOPMENT

	Speech and Language	Social Cognition	Intelligence
Down Syndrome  Chromosome 21	☹ weakness in **expressive language** (e.g. delayed transform babbling to meaningful speech)[1] ,**morphosyntactic processing** (e.g. reflexives, sentence voice) [2] , **referential communication** [3]	☹ over-friendly, hyper social weakness in **facial recognition** (e.g. recognizing facial expression) [4] , **performing theory of mind tasks** [5]	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Williams Syndrome  Chromosome 7	☺ **preserved language ability**	☹ over-friendly, hyper-social, high interest in social interaction weakness in **performing theory of mind tasks** (e.g.perspective taking) [6] , No difficulties in facial recognition [7], good superficial dyadic interaction but deficits in triadic interaction [8]	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Prader-Willis Syndrome  Chromosome 15	☹ Great **individual variety**: from **non-verbal** to **normal speech and language skills** **Speech Impairment** (e.g. reduced oral motor skills, imprecise articulation, hyper nasality, flat intonation patterns, abnormal pitch) [9]	☹ Difficulties in interpersonal relationships weakness in **facial recognition** (e.g. reading facial expression of emotions) [10] difficulties in interpreting **visual social information** (e.g poor performance on The Social Attribution Task) [11]	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Angelman Syndrome  Chromosome 15	☹ Individuals have limited expressive language ability, almost **non-verbal**. severe impairment of **receptive and expressive skills** [12]	☹ Most individuals have **hyper social** behaviors, such as frequent and spontaneous laughter. [13]	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Wolf-Hirschhorn Syndrome  Chromosome 4	☹ **Speech is usually absent** in patients with WHS. When they develop speech, they have a delayed onset and impaired development that their speech is unrecognizable, unintelligible or limited to specific sounds. [14]	☺ When individuals have expressive speech and language skills, they have good socialization abilities. [15]	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Smith-Magenis Syndrome  Chromosome 17	☹ Strong auditory memory and processing for linguistic tasks Receptive vocabulary is stronger than expressive Knowledge of word associations is better than syntactic skills [16]	☺ Individuals usually have good pragmatic skills [17]	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Turner Syndrome  X Chromosome	☺ **Generally good in language skills**	☺ **Showing no deficits in social cognition**	☺ normal Intelligence no Intellectual disabilities
Fragile X Syndrome  X Chromosome	☺ Expressive, receptive and pragmatic language abilities and literacy skills are similar to typical developed peers [18]	☹ **ASD** is a common comorbid condition of Fragile X Syndrome. Individuals with Fragile X Syndrome usually show **social avoidance** and **social indifference**. They also have deficits in recognizing **facial expression**.	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability
Cri du chat Syndrome  Chromosome 5	☹ Most individuals with Cri du chat Syndrome have **difficulties with language**.Half of the children learn sufficient verbal skill to communicate. Some individuals learn to use short sentences, while others express themselves with a few words, gestures or sign language.	☺ Both children and adults with this syndrome are usually friendly and enjoy social interactions.	☹ mild (IQ: 50-69) to moderate (IQ: 35-50) intellectual disability