

Chapter 10

Autism Genetics: What Do We Know?

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ABSTRACT

With autism there are so many questions. Researchers, educators, family members, and individuals with autism ask questions about the disorder. What causes the disorder? What are the outcomes? What does an autism diagnosis mean in terms of quality of life? Along with these practical questions, most individuals and families will also ask, “Why?” Why does one person have autism and not another? We are fortunate to be living in the 21st century where many elite researchers have developed answers to these questions and even answers, “Why?” In fact, many researchers are beginning to identify that there is a genetic component to autism. The following chapter will discuss a brief overview of critical historical research studies illuminating the relationship between autism and genetics.

INTRODUCTION

With autism, there are so many questions. What causes the disorder? What are the outcomes? What does an autism diagnosis mean in terms of quality of life? Along with these practical questions, many individuals and families will also ask “why.” Why does one person have autism and not another? We are fortunate to be living in the twenty first century where many elite researchers are developing answers to

DOI: 10.4018/978-1-7998-7732-5.ch010

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all of these questions and most significant, data is beginning to support answers to “why.” With decades of exploration researchers are collectively identifying that there is a genetic component to autism. The following chapter will discuss a brief overview of critical historical research studies illuminating the relationship between autism and genetics.

AUTISM RESEARCH HISTORY

1877 Dr. John Langdon Down was the first to describe Down’s Syndrome as he researched mental retardation. He identified three categories of mental retardation. First category was, congenital, in which there were hereditary factors that were present in diagnosis and this was the largest category. The second category was “accidental” in which injury or disease occurred late in the pregnancy or shortly after birth. The third category was “developmental” and these individuals had symptoms unlike the previous two categories, symptoms similar to what would be labeled as autistic today.

The term autism was first used by the psychiatrist Eugen Bleuler in 1908. Dr. Bleuler (1857-1939), a psychiatrist and professor, was passionate about his work, he lived with his patients, taking care of them and writing down observations (Stotz-Inenlath, 2000). Dr. Bleuler first used the term “schizophrenia” in a Berlin lecture in 1908 to describe patients who had withdrawn into their own world (Mandal, 2019). Bleuler identified deficits in these patients, specifically deficits in impaired associations, disordered affectivity and marked ambivalence (characteristics that might be considered autism today). These characteristics combed as the main symptoms for schizophrenia (Stotz-Ingenlath, 2000). But unlike other researchers of the time, Bleuler, did not believe the overall prognosis was uniformly grim. “Bleuler’s main merit is stressing on an idiographic “understanding” of the patient and plausible and subtle explanation of schizophrenia which helped to reduce the alientation of the affective persons,” (Stotz-Ingenlath, 2000, p. 1). Bleuler identified some conclusions like, the schizophrenic gene was not the dominant trait, the disease involves a complex gene dihybrid that does not include a monohybrid gene and that the disease may be a polymorphic, meaning it presents itself in many different forms. In fact, several elements likely had to come together for the disease to occur.

Bleuler began to use the term “autistic” in 1911 specifically, to describe patients with symptoms having a withdrawn from reality (Iannelli, 2020). Bleuler developed the word from the Greek word “autos” meaning self and he modified “autos” to “autism” to mean morbid self-admiration and withdrawal within self. The study of autism originated in the 1940s with the work of Dr. Leo Kanner in the US and Dr. Hans Asperger in Austria. The two leaders in research were working independently,

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