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REVIEW ARTICLE

Understanding Disorders: Insights into Phenotypes, Endophenotypes, Neurotypes, and Genotypes within the Context of the PENG Model

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ABSTRACT

Advancements in neuroscience, genetics, and psychology have paved the way for a deeper understanding of human disorders and syndromes through the study of Phenotypes, Endophenotypes, Neurotypes, and Genotypes, collectively known as the PENG model. The framework of PENG provides critical insights into the complex interplay between biological, neurological, and behavioral factors, allowing for a more comprehensive understanding of individual and group differences. This approach enhances precision in diagnostics and fosters personalized treatment interventions, ultimately contributing to better clinical outcomes. As research continues to uncover these connections, it supports a shift towards more inclusive and compassionate approaches to mental health and neurodiversity, promising new pathways for individualized care and societal acceptance.

Keywords: *Endophenotypes, Genotypes, Neurotypes, PENG model, Phenotypes*

1. INTRODUCTION

In recent years, advancements in neuroscience, genetics, and psychology have enabled a deeper exploration into the complexities of human disorders and syndromes (Grezenko et al., 2023). Previous as well as recent studies examining the relationships among phenotypes (Bieber et al., 2017; Lenzenweger, 2013; Wojczynski & Tiwari, 2008) and endophenotypes (Bieber et al., 2017; Kendler &

Neale, 2010; Lenzenweger, 2013), neurotypes and neurodiversity (Dias, Schneider, & Bohrer, 2024; Sheppard, Webb, & Wilkinson, 2024; Stones, 2023), and genotypes and epigenotypes (Karmiloff-Smith, Scerif, & Thomas, 2002; Li et al., 2023; Te Pas et al., 2017), researchers and clinicians can better understand how various biological, neurological, and behavioral factors interplay to shape not only individual and group differences, but also in animals (e.g., Kuffler et al., 2024) and plants (e.g., Li et al., 2023). These insights help illuminate the underlying mechanisms of many conditions, offering the potential for more precise diagnostics, treatments, and interventions tailored to individual needs.

Generally, educational therapists working with students with varied learning and behavioral challenges often rely their understanding on the key brain-related conceptual types (known as BRCT in short) to provide themselves an insight into how students learn, process information, and develop skills (also see Plack, 2024). The author of this paper has identified seven main BRCT: The first type has to do with the Cognitive Processing (CoP), which refers to specific mental functions (Thatcher & John, 2021), such as attention, memory, perception, language processing, and executive functions (e.g., planning, problem-solving, organization). Hence, different cognitive profiles can impact how students process information, respond to instructions, and retain knowledge (see Thatcher & John, 2021). The second type refers to the Learning Styles and Preferences (LSP; see Newton & Salvi, 2020)). Although traditional learning styles (i.e., visual, auditory, kinesthetic, and a mix of them) have mixed support, it remains useful to understand how students prefer to engage with material. Some students may be more verbal, others spatial, and some may respond better to multisensory approaches. However, in a recent study done by Rogowsky, Calhoun, and Tallal (2020) found that providing instruction based on students' learning style preferences did not improve learning. The third type concerns Emotional and Social Processing (ESP; seeVan Kleef & Côté, 2022). Emotional regulation and social cognition are always crucial, as they affect a student's ability to work in group settings, manage frustration, and navigate social interactions. For example, students struggling with emotional regulation may find it harder to focus, especially when frustrated or anxious. The fourth type is Sensory Processing (SP; Delgado-Lobete et al., 2020). Some students have sensory processing differences, such as being over- or under-sensitive to sensory input. This can influence focus and comfort in learning environments. An educational therapist may need to adjust the sensory environment to support these students. The fifth type refers to the Developmental Profiles (DP; see Brown, Parikh, & Patel, 2020). Educational therapists are quite familiar with developmental profiles, such as language, motor, and cognitive developmental stages, which offer them a better understanding of age-appropriate skills in order for them to address gaps or delays, especially with younger students or those with developmental differences.The sixth type concerns the Executive Functioning (EF; see Doebel, 2020), which involves skills like planning, working memory, inhibition, and cognitive flexibility. A student's executive function profile can affect their ability to organize, complete tasks, and adapt to changing expectations. The seventh and last type concerns the Attention (AT; see Niu, Zhong, & Yu, 2021). Different types of attention, such as sustained, selective, alternating, and divided attention, affect a student's ability to focus on tasks. This last type is often used in the task behavior analysis (TBA) to better understand how and why a student does a task in a certain way within a given context and time (Chia & Lim, 2016). Knowing these types can help educational therapists provide strategies to improve attention or manage challenges associated with it.

Understanding these aspects of the BRCT-based function enables educational therapists to design tailored intervention programs, helping students to build on strengths and compensate for areas of need. While the BRCT model is commonly known and applied, there is yet another more scientific and evidence-based model: the PENG model (Chia, 2024; Merlion Paediatric Therapy Clinic/MPTC, 2024).

1.1 The PENG Model

According to MPTC (2024), the diagnostic evaluation of an individual's condition of a disorder or syndrome is best based on the integration of data collected from following types that constitute the PENG model:

1. **P**henotype that consists of the four categories of symptoms: core, correlated, secondary and artefactual (Cassidy & Morris, 2002; Pennington, 1991; Wojczynski & Tiwari, 2008);
2. **E**ndophenotype which is based on traits linking to genetic factors not directly observable (Gottesman & Gould, 2003; ; Kendler, & Neale, 2010);
3. **N**eurotype that includes the neurological soft signs to determine overlapping conditions of a disorder (Rudy, 2023; Valeur, 2023); and
4. **G**enotype concerns the genetic composition of an individual (Chauhan, 2019; Johannsen, 1911, 2014).

As already mentioned at the beginning of this paper, phenotype goes best along with endophenotype, neurotype is better understood in the context of neurodiversity, and genotype should go with epigenotype, in order that a comprehensive assessment profile of a client can be obtained. This is, of course, an ideal case because in a real situation, such a diagnostic evaluation can be very costly and also time-consuming, as the process involves a multidisciplinary team of specialists to work collaboratively together by sharing information. Figure 1 below shows a holistic approach to diagnostic evaluation adopted by Merlion Paediatric Therapy Clinic, Singapore, in assessing, profiling and evaluating an individual condition. This is the PENG model as put forth by Chia (2024).

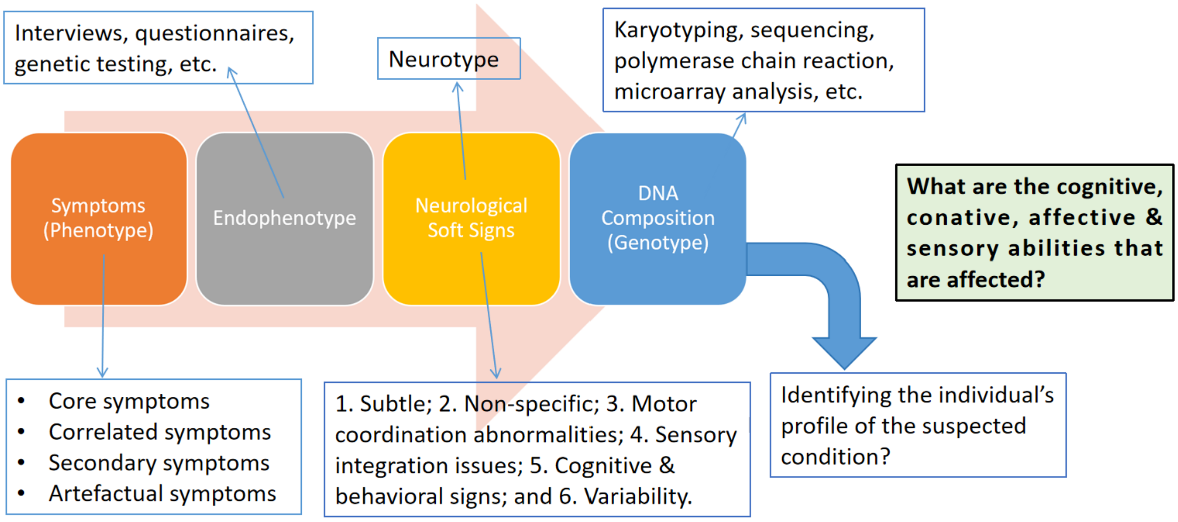

Figure 1. The PENG Model

Phenotype, endophenotype, neurotype, and genotype (PENG) each describe different aspects of biological and psychological characteristics. While they can be influenced by ecological and biological factors, their definitions and implications vary in terms of each relationship with ecological and biological contexts (see Table 1).

Table 1. PENG Definitions and Eco-Biological Bases

Type	Definition	Eco-Biological Basis
Phenotype Wojczynski & Tiwari, 2008)	The phenotype refers to the observable characteristics or traits of an organism, which can include physical attributes (morphology, physiology) as well as behavioral traits.	Phenotypes result from the interaction of an organism's genotype (its genetic makeup) and environmental influences. This means ecological factors such as climate, diet, and habitat can significantly impact the expression of phenotypic traits. For example, two plants of the same species may grow differently in varying soil conditions.
Endophenotype (Bieber et al., 2017)	An endophenotype is a genetic substructure that lies between the genotype and the phenotype, often referring to measurable traits that are	Endophenotypes can be influenced by both genetic and environmental factors, but they are typically studied in the context of diseases, especially in psychiatric and neurobiological

	thought to be more closely linked to genetic predispositions than complex phenotypic traits.	research. The expression of endophenotypes can reflect underlying biological processes that may be impacted by ecological factors, such as stressors in an environment that could affect neurodevelopment.
Neurotype (Stones, 2023)	Neurotype refers to the specific neurological as well as psychological characteristics of an individual, which can encompass variations in brain structure, function, and behavior.	Neurotypes can be shaped by both genetic predispositions and environmental factors. Factors such as nutrition, social environment, and exposure to toxins can influence neurodevelopment and lead to variations in neurotypes. For instance, certain neurodevelopmental disorders may have both genetic and environmental risk factors.
Genotype (Johannsen, 2014)	The genotype is the genetic constitution of an organism, encompassing all the genes that it possesses.	While the genotype itself is purely genetic, the expression of specific genes (phenotype) can be affected by environmental factors. Additionally, natural selection and evolutionary processes can shape genotypes based on ecological pressures, leading to adaptations that enhance survival and reproduction in specific environments.

All four concepts as described in Table 1 are interconnected and can be influenced by ecological and biological factors. The genotype provides the genetic framework, while phenotypes and endophenotypes represent the expression and intermediate traits influenced by both genetics and the environment. Neurotypes similarly integrate genetic and ecological influences but focus on neurological aspects. In short, understanding these concepts requires a consideration of both genetic and environmental contexts, making them inherently eco-biologically relevant.

2. PHENOTYPE

The Cattell-Horn-Carroll (CHC) theory of cognitive abilities (Schneider & McGrew, 2018) is primarily associated with phenotype (Tan, 2024). Phenotypes encompass observable characteristics or traits (Wojczynski & Tiwari, 2008), which in the CHC model includes measurable cognitive abilities. These traits can be classified under four categories of symptoms, i.e., core, correlated, secondary and artefactual (Pennington, 1991). The CHC theory categorizes intelligence into broad and narrow abilities that can be assessed through various cognitive tasks, reflecting phenotypic variation in intelligence (Chia, 2024).

Phenotype aside, the other three terms - endophenotype, neurotype and genotype - they are not linked to the CHC theory. For instance, the endophenotype (intermediate traits linking genes to behavior) that is often used in psychological and psychiatric research, but the CHC theory focuses on observable abilities rather than these intermediary genetic traits. The neurotype (neurological makeup or pattern) is sometimes used in discussions about neurodiversity, and it is important to note that the CHC theory does not specifically address neurotype but focuses on cognitive skills and abilities. Lastly, the genotype (genetic makeup), though the genes might influence cognitive abilities, the CHC theory itself does not address genetic information directly. In summary, the CHC theory fits within phenotypic characterization because it deals with measurable, observable cognitive abilities.

2.1 Reasons why the CHC Model of Intelligence is Phenotypic

The CHC framework of intelligence is considered phenotypic because it describes observable cognitive abilities rather than underlying genetic or neurological mechanisms. There are several reasons to support it phenotypic features: Firstly, the focus of the CHC theory is on observable traits. The CHC framework categorizes intelligence into broad and narrow abilities based on observed behaviors and cognitive performances, such as memory, reasoning, and processing speed (Schneider & McGrew, 2018). These abilities are measured through tests and assessments that capture performance outcomes rather than biological or genetic data. Next, the data collected from psychometric testing indicates that the development of the CHC model has relied heavily on factor analysis from large datasets of psychometric test scores (Jewsbury, Bowden, & Duff, 2017). Since these scores reflect measurable behavior and performance in specific tasks, the model primarily maps out phenotypic expressions of intelligence. Thirdly, there is no direct reference of the CHC theory to genetics. The CHC model does not attempt to describe the genetic or neurological basis of intelligence. Instead, it organizes cognitive abilities based on patterns observed across individuals, without attributing these patterns to specific genetic or neurobiological factors. Lastly, the CHC theory is applicable across environments and contexts (McGrew, 2009) but there are others such as McGill and Dombrowski (2019) have cautioned "several potential limitations in the CHC literature that may be responsible for this discrepancy" (p. 216). The phenotypic CHC model focuses on traits that can be observed in various settings. The CHC framework categorizes intelligence in ways that are relevant across diverse environments and contexts, reflecting that it describes surface-level traits that may be influenced by environmental factors. Therefore, the CHC framework is a model based on the outward, measurable expressions of cognitive ability, fitting the definition of a phenotypic framework rather than a genetic or biological one.

2.2 The CCAS-based Subtypes within the CHC Theory

Moreover, within the phenotypic context of the CHC theory, the author of this paper has proposed a further category of four key plausible subtypes: cognotype, conotype, affectotype, and senotype – based on the Cognition-Conation-Affect-Sensation (CCAS) model (Chia, 2010). These phenotypic subtypes are not standard terms typically associated with the conceptual framework of the CHC theory, which generally categorizes cognitive abilities into broad and narrow abilities and whose focus is on mental abilities rather than these specific typologies. Table 2 below provides a further breakdown of the four CCAS-phenotypic subtypes as if they were conceptually related to or inferred from the CHC concepts.

Table 2. CCAS-Phenotypic Subtypes

Phenotypic Subtypes	Description
1. Cognotype (Cognitive Type)	A term combining "cognition" and "type" to reflect someone's cognitive patterns or mental processing characteristics could be "cognotype". This term could hypothetically relate to a person's specific cognitive style or approach to processing information, which might be loosely associated with cognitive profiles in CHC, such as fluid reasoning (Gf), comprehension-knowledge (Gc), or processing speed (Gs). Similar to how "phenotype" represents physical characteristics and "neurotype" represents neurological patterns, "cognotype" would represent an individual's unique cognitive processing style or cognitive profile, including how they perceive, think, and process information. This could be used to study, discuss, or classify cognitive styles across individuals.
2. Conotype (Conative Type)	For a term combining "conation" (reflecting aspects of will, drive, and purposeful action) and "type", one might create something like "conotype." This term would refer to an individual's characteristic pattern of will, motivation, and intentional behavior. Like phenotype or genotype, conotype could represent the unique "motivational signature" that influences how a person approaches goals, takes initiative, and perseveres in tasks. In other words, the term could imply a behavioral or conscientiousness profile, possibly linked to non-cognitive factors that influence performance but are not direct cognitive abilities. CHC theory does not explicitly categorize personality traits like conscientiousness.
3. Affectotype (Affective Type)	A good term for combining "affect" (emotions, moods, and their patterns) and

	"type" (a classification or characteristic) could be "affectotype." This non-standard term may be related to emotional processing or affective characteristics (Gei). Although emotions can influence cognitive functioning, they are not directly a part of CHC structure of cognitive abilities, as it focuses on mental abilities like memory (Gsm), visual-spatial processing (Gv), and more. Hence, the novel term "affectotype" would refer to the characteristic patterns or styles of emotional expression, regulation, or mood that are unique to an individual or group, similar to how "phenotype" describes observable characteristics. In the version 2.5 of the CHC periodic table of human abilities, Schneider and McGrew (2018) have included emotional intelligence (Gei) with four narrow abilities in the model.
4. Senotype (Sensory Type)	This non-standard term might be interpreted as sensory processing preferences or abilities, but CHC theory does not include sensory processing as a core component of cognitive ability, but the abilities of the various senses. It combines senso- (from "sensation" or "sensory") with the -type suffix, in line with terms like phenotype and neurotype. This term could represent an individual's unique sensory processing characteristics, encompassing how they perceive and respond to sensory stimuli across modalities. Different sensotypes, such as visuo-sensotype for sight (Gv), audio-sensotype for hearing (Ga), hapto-sensotype for touch (Gh), olfacto-sensotype for smell (Go), and gusto-sensotype for taste (no specific CHC-based broad ability) as well as kinetiko-senotype for movement (Gk) might describe those who are highly sensitive to certain stimuli (like sound or touch) versus those who are less responsive, helping to capture the diversity of sensory processing experiences.

In other words, while these phenotypic subtypes could describe individual differences in broader human functioning, they do not always fit directly into the cognitive phenotype categories in CHC theory, which is structured more around measurable cognitive abilities and intellectual functions.

3. NEUROTYPE

"Neurotype" and "neurologicotype" (or "neurological type") are terms used to describe variations in brain functioning, though they are used in different contexts and with slightly different meanings. The term "neurotype" is often used in discussions around neurodiversity, referring to variations in cognitive functioning (Rudy, 2023). A person's neurotype may include traits associated with neurodevelopmental conditions, such as autism or ADHD, or it may describe a more typical (neurotypical) cognitive profile. In this context, "neurotype" helps highlight the spectrum of cognitive diversity without framing it as a medical issue. The other term "neurologicotype" is less commonly used and is a coined word to refer to structural or functional variations in the nervous system that may or may not correlate with different behavioral or cognitive traits. It is sometimes used in medical or clinical contexts to describe broader neurological differences, often with more emphasis on clinical assessment and structural aspects of the nervous system.

Neurologicotype, or sometimes referred to as "neurological type," is a concept that categorizes individuals based on the functional patterns of their nervous system. In other words, the term "neurological", according to Merriam-Wbster (n.d.), refers to "of, relating to, or affecting the nervous system" (para. 1). It explores the different ways people process information, react to stimuli, and engage with the world, often linking these patterns to brain structure, neurotransmitter activity, and neurological responses. This concept is especially relevant in understanding conditions like ADHD, autism, and sensory processing disorders, which are characterized by unique neurologicotypes that influence behavior and cognitive function. For instance, people with a highly sensitive neurologicotype might experience overstimulation in busy environments, while those with a more regulated neurologicotype might navigate similar spaces without issue.

In a broader sense, neurologicotype is used to recognize and validate diverse neurological differences, supporting the idea that variations in neurological function are part of natural human diversity rather than deficits.

In short, while "neurotype" tends to be more widely recognized in discussions of neurodiversity and cognitive differences, "neurologicotype" (or “neurological type”) may be used in medical or clinical contexts to describe broader, more structural aspects of neurological differences.

4. CONCLUSION

Understanding disorders and syndromes through the lenses of phenotypes, endophenotypes, neurotypes, and genotypes opens new avenues for personalized medicine and more compassionate approaches to mental health and neurodiversity. By identifying these intricate connections, researchers can contribute to a more nuanced view of human diversity, enhancing both clinical outcomes and societal acceptance of individuals with unique neurological and genetic makeups. As this research continues to evolve, it holds promise for future advancements in healthcare and our understanding of the human mind.

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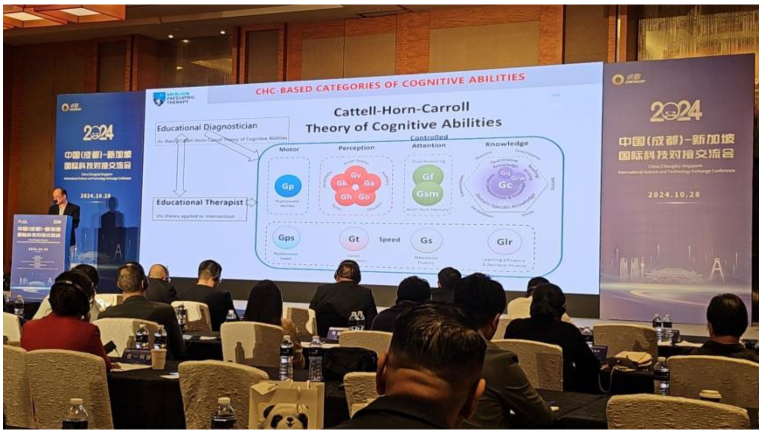
Below is a photograph taken at the conference where the author presented his paper on the CHC-based cognitive abilities in the context of the PENG model.



6. COMPETING INTERESTS

The author has declared that no competing interests exist.

7. FINANCIAL DISCLOSURE

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8. ARTIFICIAL INTELLIGENCE DISCLOSURE

No generative AI or AI-assisted technologies were used in the preparation of this manuscript.

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