Advancing Neurological Disease Treatment through the Integration of Precision Medicine and Neurogenetics

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Abstract

Neurogenetics, at the intersection of neurobiology and genetics, plays a crucial role in understanding the complex biological foundations of neurological disorders. As our knowledge of the genetic underpinnings of these conditions advances, the potential for personalized treatments grows significantly. This abstract explores the synergistic relationship between precision medicine and neurogenetics, emphasizing their transformative potential for treating a broad range of neurological diseases. Genetic factors play a key role in the onset, progression, and manifestation of disorders such as neuromuscular diseases, epilepsy, and neurodegenerative conditions like Parkinson's and Alzheimer's. Recent advancements in genomic technologies have enabled researchers to identify specific genetic variations linked to these diseases, offering a more detailed understanding of the genetic network influencing brain function. The integration of precision medicine with neurogenetics is revolutionizing therapeutic approaches. By leveraging an individual's genetic profile, precision therapies tailor interventions to target the root causes of neurological conditions, offering a more effective alternative to conventional treatments, which often have limited success due to the heterogeneous nature of these disorders. Through the exploration of genetic complexities, new therapeutic targets are emerging, and innovative treatments, including gene therapies and customized medications, are being developed. This approach not only enhances treatment efficacy but also minimizes side effects, shifting the focus toward personalized, patient-centered care. In conclusion, the fusion of precision medicine and neurogenetics is poised to open a new chapter in the management of neurological diseases. With ongoing technological advancements and a deeper understanding of genetics, the potential to improve patient outcomes and revolutionize treatment approaches by tailoring therapies to individual genetic profiles is vast.

Keywords: Neurogenetics,. Precision treatments, Neurological disorders, Genetic factors, Personalized interventions.

Significance | The convergence of precision medicine and neurogenetics revolutionizes personalized care, offering targeted, effective treatments for diverse neurological disorders.

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1. Introduction

Advancements in genetic and -omics technologies have significantly enhanced the early diagnosis of rare neurogenetic disorders, including neurodevelopmental disorders (NDDs) and inherited metabolic disorders (IMDs) (Adhikari et al., 2020). Early genetic detection is critical for developing tailored therapies that can modify disease progression and improve the quality of life for affected individuals (Annemans et al., 2017). NDDs, which directly affect 3% of the population and indirectly impact another 5% through familial caregiving and recurrence concerns, represent a widespread challenge. Patients with NDDs often experience severe

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somatic and neuropsychiatric comorbidities, such as intellectual disabilities (ID), epilepsy, behavioral and cognitive impairments, sensory deficits, and multi-organ dysfunction. These complex and lifelong care needs pose significant challenges for healthcare providers and systems striving to deliver optimal, individualized care.

Opportunities for symptomatic and disease-modifying interventions for NDDs, and even more so for IMDs, are rapidly expanding. These include dietary regimens, repurposed pharmaceuticals, organ and stem cell transplants, and emerging RNA- and gene-based therapies (Antonarakis et al., 2020). However, clinical trials for these rare patient populations face unique challenges, including methodological complexities and difficulties in establishing robust outcome metrics. Additional barriers to implementing personalized medicine arise from organizational, financial, and regulatory constraints. Translating innovative therapies into routine patient care while ensuring equitable access and reimbursement remains a time-intensive and often unsuccessful endeavor. This creates a critical gap where patients are deprived of novel treatments, and evidence-based care strategies remain underdeveloped.

To address these challenges, we review the current landscape of personalized medicine for IMDs by analyzing success stories that highlight potential improvements in trial design, outcome measures, and collaborative efforts. Furthermore, we propose leveraging the "flywheel model" (Figure 1) as a framework to accelerate the adoption of tailored therapies. This model emphasizes the central role of patients and their families within the rare disease research and care ecosystem, fostering collaboration among basic scientists, clinical experts, and ethicists. At the Emma Center for Personalized Medicine at Amsterdam UMC, this approach is applied to ensure no rare disease patient is left behind, setting a foundation for extending personalized medicine advancements to NDDs and beyond. (Barendsen et al., 2020)

2. Advances in Diagnostics

Significant progress has been made in the diagnosis of individuals with rare diseases, including presymptomatic individuals, children, and adults with developmental delay, epilepsy, neurocognitive decline, or other complex disorders. These advancements also benefit severely ill children, enabling faster diagnoses and minimizing the "diagnostic odyssey" that patients and their families often endure. Rapid and accurate diagnosis not only provides a clearer prognosis and recurrence risk counseling but also supports caregivers in accepting their condition. It facilitates connections with patient organizations for peer support and fosters engagement with personalized health monitoring and treatments, which can potentially reverse disease progression (Table 1).

Population-based genetic screening has expanded the opportunities for early detection of rare diseases before clinical manifestation. While genetic testing was previously considered time-intensive, the advent of next-generation sequencing (NGS) has transformed the landscape. NGS has shown the potential to provide diagnoses and treatment possibilities in approximately 73% of families with acutely ill children (Bianchi & Vai, 2019). Screening strategies range from preconception and preimplantation carrier screening in parents to prenatal and neonatal programs for offspring. NGS has also revolutionized the identification of novel gene variants associated with rare Mendelian diseases (Blau et al., 2014).

Exome sequencing, a key application of NGS, has been instrumental in directly influencing care in 15.6% of cases and in as many as 39% of rare pediatric diseases (Bok et al., 2012). For certain conditions, the diagnostic yield can reach 41%. Studies in adults with epilepsy, autism spectrum disorder, and intellectual disability (ID) report similar findings (Boycott & Ardigó, 2018). Whole-exome sequencing (WES), combined with clinical and biochemical phenotyping, has achieved a 68% diagnostic rate in children with developmental delays, with 44% of cases requiring adjustments to metabolic treatments (Boycott et al., 2020).

Despite the effectiveness of WES, a molecular diagnosis remains elusive for at least one-third of patients with rare disease phenotypes. Several factors contribute to this "missing heritability," including technical limitations (e.g., structural variants, intronic mutations, and coding variants missed by WES), complex biological mechanisms (e.g., somatic mosaicism, allelic expression imbalances, and tandem repeat expansions), and polygenic inheritance observed in 4–9% of rare disease cases (Budimirovic et al., 2017).

Whole-genome sequencing (WGS) offers a more sensitive approach for detecting specific genetic variations, such as copy-number variants (CNVs), chromosomal rearrangements, and regulatory-region mutations, that may be missed by WES. Paired with RNA sequencing, WGS can be a powerful tool for resolving unsolved inherited metabolic disorders (IMDs) (Cakici et al., 2020). Splice-site variants and their associated biological pathways are increasingly being targeted with RNA-based therapies (Camfield & Camfield, 2011). However, interpreting variants of uncertain significance, addressing incidental findings, and navigating societal stigma remain critical challenges in implementing these technologies (Centerwall & Centerwall, 2000). Nevertheless, parents of affected children exhibit high acceptance of genetic findings, with minimal regrets even when faced with these complexities (Coppus, 2013).

In addition to genomic approaches, advancements in other "-omics" technologies are transforming the diagnostic landscape. Methylomics, for example, provides genome-wide methylation analyses that identify biological markers and support the validation

of disease-causing genes. Methylation profiling has uncovered diagnostic markers for over 48 neurological disorders, particularly those involving gene mutations that affect methylation states (Coughlin II et al., 2015).

Metabolomics, lipidomics, and glycomics enable the simultaneous profiling of thousands of metabolites, lipids, and glycans in biological samples, facilitating the identification of traits missed by genomic approaches (Coughlin et al., 2021). These technologies, when combined with model organisms, offer potential for exploring novel phenotypes. Integrating these "systems biology" methods requires robust bioinformatics and collaborative efforts between molecular geneticists, IMD clinicians, laboratory specialists, and researchers. In translational metabolic research, techniques like gene expression analysis, CRISPR–Cas technology, and genetic engineering are employed to unravel disease mechanisms, identify biomarkers, and develop therapeutic interventions (Demos et al., 2019).

This multiomics approach has yielded significant diagnostic and therapeutic advancements. For example, a study by (Den Hollander et al., 2021) identified 11 new disease genes and over 20 novel phenotypes. The study diagnosed 90% of 41 families with unexplained neurometabolic phenotypes, demonstrating how combined phenomics and multiomics approaches can provide pathophysiological insights. This led to optimized treatment strategies in over 40% of cases, in addition to increasing diagnostic yield.

One notable example is the discovery of NANS-congenital disorder of glycosylation (NANS-CDG), a newly identified IMD characterized by impaired sialic acid biosynthesis. This syndrome, identified in nine patients with ID and skeletal abnormalities, highlights the value of untargeted metabolomics over WES and phenomics (Eichler et al., 2017). The identification of N-acetyl-mannosamine as a biomarker in Dutch patients underscored its diagnostic significance. Subsequent deep phenotyping studies revealed a genotype-phenotype correlation and characteristic MRI abnormalities associated with NANS-CDG (Engelke et al., 2021). Preclinical studies using zebrafish models demonstrated that sialic acid supplementation could correct the phenotype, leading to human clinical trials and induced pluripotent stem cell (iPSC) research to explore therapeutic options.

In conclusion, advances in diagnostics, particularly through genomic and multiomics technologies, have transformed the landscape of rare disease research and patient care. Early and accurate diagnosis not only alleviates diagnostic delays but also paves the way for targeted treatments and improved outcomes. Continued innovation in this field, coupled with collaborative efforts, holds the promise of unlocking new therapeutic opportunities and enhancing the lives of patients with rare diseases.

Predictive Medicine: Phenotypic Modifiers and Prognosis

To optimize prognosis and determine the timing of interventions, it is essential to understand the factors influencing disease progression. A striking example is X-linked adrenoleukodystrophy (ALD), where prognosis cannot be reliably predicted at diagnosis. However, timely hematopoietic stem cell transplantation (HSCT) can prevent severe and often fatal central nervous system deterioration (Falkenberg et al., 2017). Early brain changes in a small group of affected males can only be detected through frequent brain MRI scans. Interestingly, phenotypic discordance in siblings and monozygotic twins suggests that additional (epi-)genetic modifiers may trigger the demyelination process.

Recent research using multi-omics approaches identified different phenotypic modifiers in six brothers with discordant early and late disease onset, offering new insights into ALD progression (Ferreira et al., 2019). With the inclusion of ALD in newborn screening programs, further studies are needed to better understand these modifiers and their role in disease progression (Ferreira et al., 2021). Gene therapy is also emerging as a potential treatment for this devastating condition (Kotulska et al., 2021). Moreover, insights into protective modifiers may pave the way for developing new preventive measures and treatments that enhance resilience.

3. From Diagnosis to Treatment

Early diagnosis, ideally during the critical neurodevelopmental window, offers the opportunity to apply disease-modifying therapies and potentially prevent somatic complications, autism, or intellectual disabilities (Kremer et al., 2017). Advances at the genomic, epigenomic-transcriptomic, and metabolomic levels have identified promising therapeutic targets. Model systems such as organisms, organoids, and induced pluripotent stem cells (iPSCs) enable deep phenotyping to uncover biomarkers. Complementary approaches—including radiological, electrophysiological, hematological, somatic, and neuropsychiatric characterization—further enhance the identification of therapeutic opportunities.

Figure 1 illustrates the interconnected processes critical for treatment success, represented by the flywheel's various blades. Progress in this challenging field necessitates collaboration within specialized personalized medicine facilities. These centers integrate resources and expertise to assess the impact of interventions—such as diet, supplements, repurposed drugs, transplantation, and RNA or gene therapy—on relevant outcomes. These evaluations employ cutting-edge -omics technologies and cell models in vitro (Krueger et al., 2010) and are validated in vivo through clinical trials tailored for rare diseases (Lee et al., 2018).

4. Inherited Metabolic Disorders as a Model

Inherited metabolic disorders (IMDs) exemplify how advances in understanding genetic and biochemical deficiencies over the past

century have led to the development of targeted treatments. While individually rare, IMDs collectively affect approximately 1 in 800 newborns, representing a significant global health burden. These monogenic disorders often result from a missing enzyme or transporter, leading to metabolic blockages, substrate accumulation, and deficits in essential building blocks. These disruptions can progressively damage organs such as the kidneys, liver, brain, heart, or eyes. With more than 1,600 known IMDs, this group constitutes the largest category of monogenic disorders for which targeted therapies—such as enzyme replacement, gene/RNA therapy, dietary modification, bone marrow transplantation, and nutritional supplements—are either under development or already available (Mueller et al., 2016).IMDs are increasingly recognized as a significant cause of previously unexplained conditions like intellectual disabilities (ID). For instance, a systematic study using mass spectrometry as a first-line screening test identified IMDs as the etiology in 8% of 518 patients with ID, many of whom were treatable (Müller et al., 2021). These breakthroughs underscore the importance of understanding pathophysiological mechanisms to develop and evaluate specific therapies.

A landmark example is phenylketonuria (PKU). In 1934, Følling identified the causative deficiency in phenylalanine hydroxylase by analyzing two children with ID and a distinctive urine odor caused by elevated phenylpyruvic acid (Overwater et al., 2019). In 1954, Dr. H. Bickel successfully treated PKU using a phenylalanine-restricted diet, which laid the foundation for the heel prick newborn screening technique introduced by Guthrie in 1962. This screening program has prevented severe cognitive impairments globally. However, the lifelong dietary restrictions are burdensome, prompting the development of novel therapies. These include BH4 cofactor supplementation to enhance residual enzyme activity and gene replacement or subcutaneous enzyme replacement therapies, both of which are undergoing clinical trials. Collaboration among researchers, clinicians, and PKU patient associations has been instrumental in these advances, exemplifying P4 medicine—preventive, predictive, personalized, and participatory (Petrikin et al., 2015).

Another notable case is pyridoxine-dependent epilepsy (PDE). First described in 1951 as neonatal seizures resistant to conventional treatments but responsive to vitamin B6, PDE was later identified as a neurometabolic disorder caused by impaired lysine metabolism. This impairment leads to the accumulation of toxic metabolites, such as alpha-aminoadipic acid semialdehyde (α-AASA) (Posey et al., 2017). Despite controlling seizures with pyridoxine, 75% of individuals still experience intellectual disabilities (Quaio et al., 2020).

To address this, the International PDE Consortium was established to develop better treatments. Collaborating closely with patients and families, the consortium devised lysine reduction therapy (LRT), which combines dietary modifications with arginine supplementation to reduce α-AASA levels. This approach has improved seizure control and psychomotor development in affected individuals (Richmond et al., 2020). Further validation of LRT's efficacy led to the establishment of the PDE care pathway and international consensus standards (Rosso et al., 2020).

A critical milestone was the identification of a reliable biomarker, 2-OPP, detectable in neonatal screening bloodspot cards, enabling early diagnosis and timely treatment (). Efforts are ongoing to explore additional therapeutic options, such as upstream enzyme inhibition. PDE's progress in diagnosis, treatment, and prevention surpasses most IMDs, which often remain undiagnosed until late stages and lack curative options. Moreover, there may still be hundreds of IMDs yet to be classified or treated (Sadikovic et al., 2021).

Recent breakthroughs in non-nutraceutical IMD therapies highlight the field's potential. For example, asfotase alfa, an enzyme replacement therapy targeting tissue-nonspecific alkaline phosphatase (TNSALP), has effectively treated hypophosphatemia's debilitating bone phenotype and associated epilepsy (Sahin & Sur, 2015). Similarly, lumasiran, an RNA interference (RNAi) therapy, reduces urinary oxalate excretion in patients with primary hyperoxaluria type 1, slowing kidney failure progression (Satterstrom et al., 2020).

Personalized medicine has also reached unprecedented levels, as demonstrated by the development of milasen, a splice-site-modifying antisense oligonucleotide specifically created for Mila, a patient with CLN7 disease. This treatment was delivered within a year of diagnosis in an N-of-1 study. Although seizures initially improved, the disease ultimately proved fatal, underscoring the challenges in treating advanced IMDs (Schenkel et al., 2017).

Excitingly, gene therapies are becoming a reality. The European Medicines Agency (EMA) recently approved Libmeldy, an ex vivo gene therapy for metachromatic leukodystrophy. This therapy uses autologous CD34+ stem cells transduced with a lentiviral vector, representing a significant advance in treating rare genetic disorders (Senn, 2016).The collective progress in IMD research exemplifies the transformative potential of precision medicine. By integrating advanced -omics technologies, innovative therapies, and collaborative efforts, researchers are paving the way for improved outcomes in conditions once deemed untreatable. Through preventive, predictive, personalized, and participatory approaches, the future of IMD treatment holds immense promise for affected individuals and their families.

5. Neurogenetic Disorder Trials and Tribulations: Personalized Trial Design

Producing evidence for rare diseases such as inherited metabolic disorders (IMDs) present significant challenges due to their

Figure 1.personalized medicine for all rare disease patients

Table 1. Advances in Neurogenetic Diagnostics

Diagnostic Technology	**Discription**
Next-Generation Sequencing	High-throughput DNA sequencing for rapid identification of genetic variations.
Functional Magnetic Resonance Imaging (fMRI)	Imaging technique measuring brain activity by detecting changes in blood flow, aiding in the study of neurological conditions.
Liquid Biopsy	Non-invasive detection of genetic material in bodily fluids, providing insights into neurogenetic markers
Biomarker Analysis	Identification and quantification of specific molecules indicative of neurological disorders, aiding in early diagnosis.
CRISPR-Based Diagnostics	Utilizing CRISPR technology for highly sensitive and specific detection of genetic mutations associated with neurological disorders.

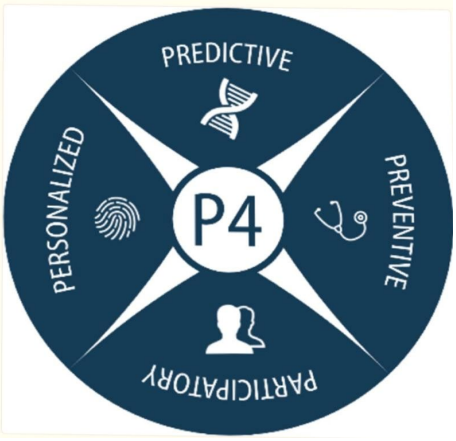

Figure 2. P4 medicine model

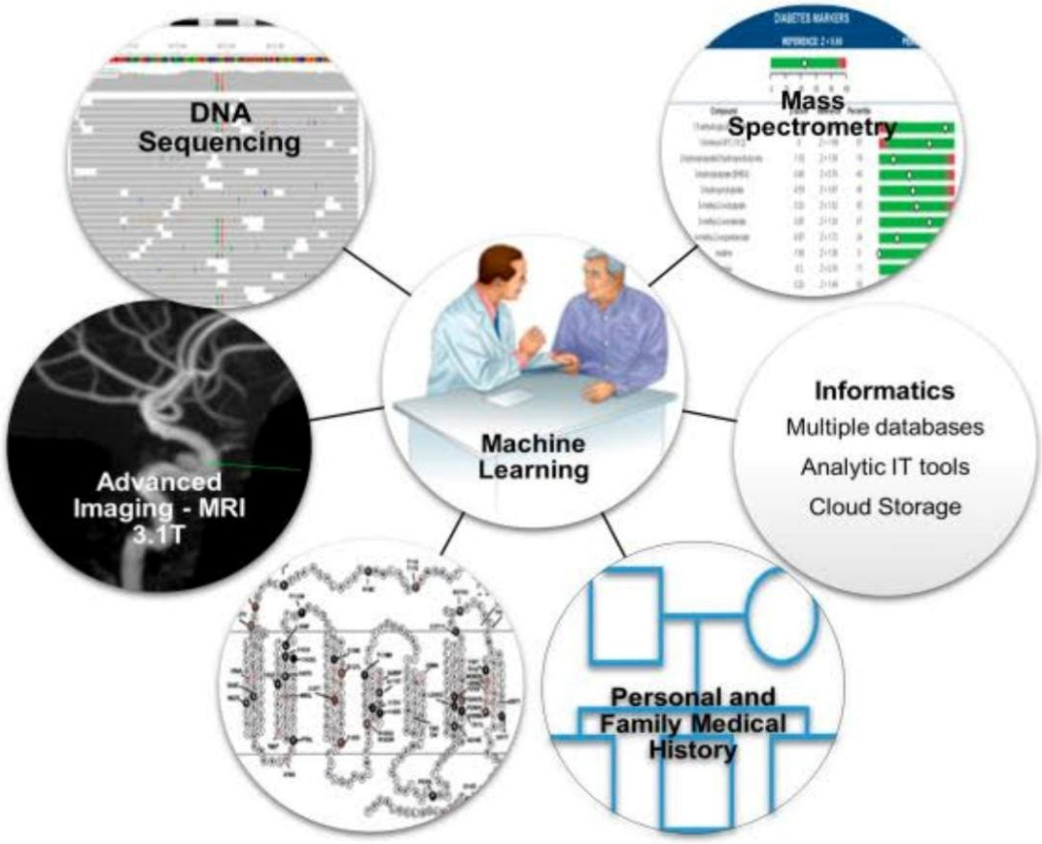

Figure 3. Personalized Treatment

heterogeneity and small patient populations (Senn, 2019). Conventional trial designs are particularly limited by the inter- and intra-individual variability of neuropsychiatric and somatic manifestations, which can fluctuate significantly over time. Additionally, randomized controlled trials (RCTs) for IMDs often exhibit wide variability in treatment responses, leading to unfavorable results for the broader group. This variability may deprive certain patients of critical treatments (Slade et al., 2018). Consequently, a paradigm shift is required for trial designs that better align with the principles of personalized medicine (Figure 3). One promising approach for rare disorders is the use of single-case experimental designs (SCEDs), such as the N-of-1 (A-B-A) design or multiple baseline design (Tarailo-Graovac et al., 2016). SCEDs meticulously assess causal relationships by conducting repeated crossover trials within a single patient under controlled, randomized conditions. Unlike traditional RCTs, which focus on group-level average treatment effects, N-of-1 studies address individual variability by identifying specific factors influencing treatment response. A series of SCEDs allows for cross-disorder comparisons and enhances the generalizability of findings to the wider patient population.

For an N-of-1 study to be effective, the targeted condition or comorbidities must remain relatively stable over time. However, many IMDs and neurogenetic disorders (NDDs) are (neuro)degenerative, making their natural course unpredictable and variable (van Karnebeek & Jaggumantri, 2015). In such cases, SCEDs can still capture the effects of interventions by monitoring baseline, placebo, and follow-up assessments to trace persistent influences on the patient's trajectory. These designs are particularly well-suited for evaluating disease-modifying therapies, which are expected to have longer-lasting effects compared to symptomatic treatments.

Establishing a tailored baseline is critical to understanding manifestations without intervention. Longitudinal monitoring and follow-ups enhance internal validity, providing robust data on the efficacy and tolerance of treatments. Patient registries enable deep phenotyping to identify (surrogate) biomarkers for tracking disease progression and therapy response. Additionally, sample size estimates guide the number of inclusions necessary for generalizing results to similar patient populations (Vissers et al., 2016). Advanced statistical approaches, such as mixed-effects and Bayesian models, further accommodate intra- and inter-patient variability, ensuring accurate assessment of treatment effects (Wagstaff et al., 2021). However, personalized trial designs such as SCEDs offer a powerful alternative to traditional RCTs for IMDs and NDDs, addressing individual variability and advancing precision medicine in rare disorders.

6. Personalized Outcome Measures

Patients with inherited metabolic disorders (IMDs) face significant challenges due to severe comorbidities and complex contextual factors. Personalized, disorder-specific outcome metrics are urgently needed to address their unique needs. These metrics should aim to enhance patient relevance and provide pathophysiological insights through minimally invasive techniques such as digital apps. Outcome measures should encompass objective biological results, validated symptom checklists, cognitive tests, and tailored metrics. The World Health Organization's International Classification of Functioning, Disability, and Health (ICF) provides a comprehensive framework for selecting outcome measures that assess the diagnosis's impact on all aspects of life and guide interventions to maximize quality of life (Wanders et al., 2019).

Quantitative tools, such as experience-sampling methods (Warmerdam et al., 2020), goal attainment scaling, and patient-reported outcome measures (Zapata-Pérez et al., 2021), effectively capture significant subjective patient experiences and transform them into evidence. For patients with intellectual disabilities (ID) who cannot articulate their clinical state, caregiver and parental support is vital. Proxy-friendly evaluation tools are essential to ensure accurate assessments and trial compliance. A patient-centered approach, involving participants in the design and evaluation of interventions, can significantly improve adherence and engagement by enhancing the perceived relevance of the trial (Cacabelos., 2017).

Unfortunately, validated and accessible outcome measures for rare NDD/IMD cohorts are often unavailable. Even in validation studies, the responsiveness of these tools to therapeutic changes is rarely assessed, a critical oversight that has previously led to disappointing trial outcomes in neurodevelopmental disorders (Kim et al., 2016). To ensure therapeutic efficacy, future research must prioritize developing responsive metrics that accurately evaluate treatment impact. This personalized approach holds the potential to revolutionize care for these vulnerable populations.

7. Discussion

The International Classification of Functioning, Disability, and Health (ICF) framework should guide counseling across all life domains following genetic diagnosis, care, and treatment planning. Ideally, this process should be integrated with the establishment and education of a local care network. Increasingly, expert centers for specific diseases and patient groups are being developed at the national level, while European Reference Networks (ERNs) such as MetabERN and ERN ITHACA foster international collaborations (Dauncey., 2013). These initiatives help build patient-friendly, transmural care networks supported by shared electronic patient records. Publicly available resources, such as the Treatable ID app

and IEMBase, facilitate easy access to diagnostic and treatment information, lowering barriers for both patients and healthcare professionals.

To ensure comprehensive care, registries—ideally patient-owned—are essential for tracking patient characteristics across all life domains and collecting longitudinal patient-reported outcomes. Such registries enable patients and healthcare providers to assess comorbidity levels and evaluate intervention effectiveness over time. They also ensure that individual patient needs are well understood. Lifelong surveillance, supported by digital tools, helps prevent the isolation of patients with intellectual disabilities (IDs) who might otherwise grow up "known well by no one" (Erro et al., 2018).

Collaboration must occur at local, national, and transnational levels. Knowledge-sharing platforms can offer guidance on trial design, outcome measures, and regulatory considerations for inherited metabolic disorders (IMDs) and neurodevelopmental disorders (NDDs). Training programs for students, academic researchers, and community physicians—featuring patient educators—are vital for understanding the multifaceted impact of IMDs and NDDs on patients and their families across ICF domains. Multidisciplinary clinical and research meetings, along with international exchange programs and fellowships, can deepen awareness of diagnostic, therapeutic, and implementation efforts. These initiatives aim to reduce health disparities and improve patient care.

The challenge remains: how can these efforts be sustainably funded? Advocacy by patient groups and healthcare professionals will continue to drive rare disease awareness at national and international funding and regulatory bodies. Validation studies can strengthen consensus on care needs, including access to orphan drugs (Campdelacreu., 2014), and further support personalized treatment approaches (Schepenjans., 2016). Platforms like European Reference Networks, Medicine for Society in the Netherlands, and the National Centers of Excellence Programs for Rare Disorders in the United States bring together academia, industry, government, and patients to ensure affordability and equitable access to rare disease therapies.

Patient preferences and interests must remain central to all efforts, with ethical, legal, and social implications (ELSI) carefully evaluated. Not all possible interventions should be implemented without thorough research and stakeholder collaboration (Wesfall., 2017). This principle applies to population-based screening programs and preventive interventions. Genomic technologies, for example, can now detect conditions undetectable by mass spectrometry, necessitating careful studies to weigh their benefits and risks. These steps are critical to achieving the second goal of the International Rare Diseases Research Consortium: the approval of 1,000 new rare disease therapies by 2027.

8. Conclusion

In conclusion, the convergence of neurogenetics and precision medicine heralds a transformative era in understanding and treating neurological disorders. The unraveling of genetic complexities underlying these conditions has provided unprecedented insights into biochemical pathways and genetic variations that influence disease manifestation. This knowledge has paved the way for tailored therapies that shift from a one-size-fits-all approach to a patient-centered paradigm, with the potential to significantly improve lives.

The integration of genomic technologies has facilitated the development of personalized treatment regimens and innovative gene therapies, offering the promise of enhanced treatment efficacy with fewer side effects. This advancement not only raises the standard of care but also redefines the approach to managing neurological conditions by addressing their root genetic causes rather than solely mitigating symptoms.

As the field continues to evolve, the identification of genetic variants is being translated into clinically actionable therapies, broadening the spectrum of treatment options. This dynamic progress underscores the potential to halt or even reverse disease progression by targeting its underlying genetic drivers.

The synergy between neurogenetics and precision medicine offers a promising future for managing neurological disorders, where treatments are tailored to the unique genetic profiles of individuals. This paradigm shift stands to revolutionize care, improve outcomes, and enhance the quality of life for patients worldwide. With ongoing research and innovation, the future of neurological medicine is poised for groundbreaking advancements, ushering in a new standard of precision-driven care.

Author contributions

S.S.K. contributed to conceptualization, methodology, and manuscript drafting. J.A.C. was responsible for supervision, critical revision of the manuscript, and project administration. W.Z. performed data collection, analysis, and interpretation. All authors read and approved the final manuscript.

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Competing financial interests

The authors have no conflict of interest.

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