

# Tourette Syndrome

## A Catalyst for Operationalizing Neurodiversity

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### Abstract

Our goal, taking Tourette syndrome as a case example, is to introduce neurologists to, and motivate discussion on, the neurodiversity paradigm. This philosophical construct considers some neurologic conditions in diversity, instead of simply disease. Moving from philosophical idea to empirical construct draws from patient and family perspectives on (1) quality of life and discrimination, (2) disability pride, and (3) unique profiles of different patient cohorts. Listening to patient voices, attending to family, advocacy group, and societal views on neurologic disorders can strengthen precision neurology practice. Dialogs on neurodiversity, including antitherapy sentiments, offer to enhance neurologic care, patient agency, and autonomy; encourage respectful communications with patients who challenge the idea their condition is pathologic; and to set the stage for future empirical investigations and practice guidelines.

### Introduction

Ari Ne’eman—an Obama appointee to the National Council on Disability—proclaimed: “If you say, ‘I want to cure... an autistic child,’ what you’re actually saying is... ‘I’m really aiming to replace that child with the child that I wish that I had.’”<sup>1</sup> Similar sentiments are expressed on autistic advocacy blogs, for example, “Asperger’s [sic] cannot be cured, nor should it be,”<sup>2</sup> and even in *Time* magazine: “We don’t need a cure for autism.”<sup>3</sup>

Antitherapy sentiments go beyond rhetoric. Spectrum 10k, a \$4 million study examining genetic and environmental correlates associated with autism spectrum disorder (ASD), was initially slated to begin in 2021. The project was, however, paused for 2 years in response to concerns that the research would be used to “‘cure’ or eradicate” ASD.<sup>4,5</sup>

Antitherapy sentiments within the autistic community are often associated with the *neurodiversity paradigm*, a neurophilosophical perspective that challenges the view that autism is pathologic. Although originally coined as a descriptor specific to autism, the neurodiversity paradigm has increasingly been promoted regarding Tourette syndrome (TS), epilepsy, dyspraxia, dyscalculia, and other neurologic disorders.<sup>6,7</sup> Neurologists may thus increasingly encounter activists hostile to cures for neurologic disorders, as well as patients and families skeptical of medical intervention. Taking TS as an exemplar, we will introduce neurologists to the neurodiversity paradigm, explore implications for research and practice, and provide a heuristic framework for neurologists, supporting operationalizable recommendations for moving forward.

Our overarching goal is not to advocate for—or against—the neurodiversity paradigm. Largely because of neurodiversity activism, autism research has been characterized as “a ‘field in crisis,’ plagued by conflict between researchers and the Autistic community.”<sup>8</sup> Neurologists and their patients will benefit if neurology can more astutely navigate the challenges posed by the neurodiversity paradigm. Yet it is unclear how disagreement over a neurophilosophical construct can be resolved. Our aim is thus to demonstrate that the neurodiversity paradigm is

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Glossary

AI = artificial intelligence; ASD = autism spectrum disorder; QOL = quality of life; TS = Tourette syndrome.

empirically tractable and, thereby, to chart a course to constructive and scientific investigation of the paradigm’s central claims because they relate to neurology. Operationalizing the neurodiversity paradigm requires a better understanding of ongoing cultural debates on the paradigm, and its potential added value for informing strategies for offering care, that honor and incorporate neurophilosophical and personal perspectives of persons living with a range of functional abilities, as in TS.

The Neurodiversity Paradigm and Antitherapy

Neurologic disorders are generally understood to be pathologic—as structural, biochemical, circuit, or electrical abnormalities in the brain, spinal cord, or peripheral nerves which generate a range of symptoms—the kind of things that make a person’s life worse.<sup>7</sup> The neurodiversity paradigm rejects this view, holding that neurologic disorders such as TS are a type of nonpathologic diversity, akin to sex or sexual orientation (Table).

The neurodiversity paradigm does not ignore the challenges of living with TS. Proponents of the neurodiversity paradigm generally hold that these challenges are best explained by discrimination or prejudice.<sup>7-9</sup> Instead of explaining TS patients’ struggles in their motor disorder, neurodiversity paradigm proponents will point to the discrimination patients with TS face because of their verbal and motor tics.<sup>10</sup>

Some of the challenges of living with TS cannot be explained in ableist bias. This alone does not offer clearcut evidence against the neurodiversity paradigm. Many differences that we do not classify as disorders involve unique challenges. For example, the range of reproductive choices available to male patients are limited by the fact that, unlike female patients, male patients are unable to conceive and carry a child.<sup>11</sup> Nonetheless, being male is not pathologic and is core to many peoples’ identities. Proponents of the neurodiversity paradigm may thus be able to accept that living with TS involves

serious challenges that are independent of ableist bias while simultaneously denying that TS should be understood as a pathology.

It is a small but critical step from accepting the neurodiversity paradigm to embracing antitherapy. If TS is like sex or sexual orientation, then attempting to “cure” or “treat” TS may be no different than so-called “Conversion Therapy,” a practice that aims to use the tools of biomedicine to *cure* being gay.<sup>12</sup> If the analogy to sex and sexual orientation holds then, just as it is wrong to try and *cure* being gay, we should think that it is similarly wrong to try and *cure* or *treat* TS. Nonetheless, though often antitherapy, neurodiversity activists are likely to embrace efforts by health care providers to improve accessibility and to design and disseminate assistive technologies.<sup>13</sup>

Classifying Neurologic Disorders: Pathologic, Neurodivergent, or Both?

Populations diagnosed with TS are unlikely to capture the divergent presentations across all persons who might qualify for TS diagnosis. Among persons diagnosed with TS, there can be profound sequelae across the lifespan. In one study, 60% of pediatric patients with TS reported suffering physical pain because of their tics.<sup>14</sup> Patients with TS or chronic tic disorders are 4 times more likely to die by suicide than control participants.<sup>15</sup> Ten percentage of children with TS report have reported attempting suicide at least once in the past year, while 27% of adults with TS report the same.<sup>16</sup> Given profound harms associated with TS, is the neurodiversity paradigm viable?

The neurodiversity paradigm also has profound implications for neurologic research and practice. If one takes the function—or *telos*—of medicine to be the treatment of disease, embracing the neurodiversity paradigm may leave TS interventions outside of the domain of medicine altogether.<sup>17</sup> More pragmatically, it may be unclear how neurologists can bill for services that do not attempt to address or remedy deficits or pathology. Similarly, if TS is viewed as merely a type of diversity, akin to sex or sexual orientation, it is unclear what would be involved in FDA approval for TS interventions, for example, what would be involved in assessing the balance of risks and *health* benefits? Embracing the neurodiversity paradigm regarding TS may either require fundamental shifts in our medical institutions or threaten to leave patients with TS outside of our medical system entirely.

A practical framework and approach is needed to better understand why patients and their families may accept the neurodiversity paradigm and to set the stage for empirical investigations of the paradigm that allow neurology to avoid

Table Disease vs Difference

Pathologic differences	Nonpathologic differences
Cancer	Ethnicity
HIV/AIDS	Sex
Alzheimer disease	Sexual orientation
Tourette syndrome (?)	Tourette syndrome (?)

the conflict that characterizes debates over neurodiversity in other disciplines. Empirical investigation of the neurodiversity paradigm can supplement the contemporary debate by leveraging inductive and abductive inferences to push beyond the analogic reasoning currently ubiquitous in the neurodiversity literature, and can further clarify problem boundaries, for example, what conditions and what severity of those conditions should be viewed as merely neurodivergent rather than pathologic?

We propose that the neurodiversity paradigm can be operationalized along 3 dimensions: (1) quality of life (QOL) and discrimination, (2) disability pride, and (3) unique strengths. That the neurodiversity paradigm can be operationalized across multiple dimensions suggests pathways and outcomes for testing empirical predictions; the neurodiversity paradigm is empirically tractable, through and through. This key result encourages more constructive and scientifically informed engagement with the neurodiversity paradigm.

Whether some aspect of TS is pathologic or neurodivergent may prove contextually sensitive to case-by-case variables in the lives of individual patients. Conditions, such as anosognosia, anosodiaphoria, amnesia, and hypochondriasis, underscore that patient testimony alone can mislead clinical decisions, including in TS. Increasingly fine-grained measures, from growing scientific discovery and emphasis on aspects of precision health (e.g., predictive, personalized, preventive, participatory, genetic, disease-specific, and environmental), will enable clearer separations between neurodivergence and disease in clinical contexts.

## QOL and Discrimination

QOL studies consistently report that patients with TS have lower QOL than their non-TS counterparts,<sup>18,19</sup> potentially indicating that, contra the neurodiversity paradigm, TS makes life worse. The connection between QOL and TS is, however, more complicated.

Among others, Chapman and Kapp argued that ableist bias is the source of the primary harms involved with many neurological disorders.<sup>10,20</sup> This understanding of TS's effect on QOL is poignantly captured by Hollenbeck observation that "Tourette Syndrome is largely a disease of the onlooker. When I tic, I am usually not the problem. You are."<sup>21</sup> If decreases in QOL associated with TS primarily result from ableist bias, QOL measures may actually provide evidence in support of the neurodiversity paradigm.

The etiology of decreases in QOL associated with TS remains unclear. Consider, for example, the GTS-QOL, a QOL measure validated for use with patients with TS.<sup>22</sup> Of the 27 measures in the GTS-QOL only one, "pain or injuries," is clearly unrelated to either interpersonal or institutional ableist bias. Of 27 validated measures, 11 seem to straightforwardly measure the effect of ableist bias, for example, "lack of social support," "difficulties seeing friends," and "embarrassing gestures."

Other measures may capture ableist bias, though less straightforwardly. For example, the GTS-QOL considers "phonic tics" and "repeating words" as physical-compulsive and obsessive-compulsive characteristics, respectively, that decrease QOL. Yet it is unclear: if phonic tics and the repeating of words were socially acceptable, would either would decrease QOL?

A third group of measures may highlight institutional, rather than interpersonal, ableist bias. For example, although patients with TS may have "difficulty concentrating" and "difficulty finishing tasks," these difficulties may reflect the demands of environments designed around the assumption of neurotypicality. For example, although the effect of exercise on tic frequency remains unclear,<sup>23</sup> traditional school and work environments are generally sedentary, which may exacerbate the symptoms of TS.<sup>24</sup>

Although patients with TS consistently have lower QOL than neurotypical peers, extant validated QOL measures do not disentangle effects on QOL intrinsic to TS and effects on QOL that result from interpersonal and institutional ableist bias. Although social stigma plays a preeminent role in the QOL decreases associated with TS,<sup>25-27</sup> the effect of institutionalized ableist bias and the effect of intrinsic features of the syndrome remain opaque. Existing QOL measures are thus too coarse grained to operationalize the claims of the neurodiversity paradigm regarding TS. Appropriately designed QOL measures are needed to better guide clinical and research decisions and to facilitate the scientific investigation of the accuracy of the neurodiversity paradigm.

## Disability Pride

Drawing on an analogy to the Gay Pride movement, Elizabeth Barnes has argued that the Disability Pride offers evidence that although (physical) disabilities can negatively affect individual lives, they are nonetheless a type of difference on par with sex or sexual orientation.<sup>11</sup> Much as the Gay Pride movement holds that nonheteronormativity is valuable and should be celebrated, the Disability Pride movement holds that (some) disabilities are valuable and should be celebrated. This view is expressed in the art, theatre, film, and parades of the Disability Pride community.<sup>11,28,29</sup>

Barnes argues that, just as we should defer to the testimony of members of the Gay Pride movement about being gay, we should similarly defer to the testimony of members of Disability Pride about being disabled.<sup>11</sup> Members of Disability Pride, not institutional medicine, have privileged insight into what life with a disability is like and, consequently, have privileged insight into the nature of disability.

Barnes' argument offers a second route for operationalizing the claims of the neurodiversity paradigm. The more TS is represented among the membership of Disability Pride, the more plausible it is to hold that TS is not pathologic.

No systematic survey of the membership of the Disability Pride movement has been completed. Pragmatic questions, like the following, remain to be answered: what qualifies as membership in Disability Pride? Must one be a member of some organization, and if so, which organizations qualify? If not, how is membership in Disability Pride measured and, without the ability to survey organizations about their membership, how would one collect the requisite data?

Theoretical questions also remain unanswered. What level of representation of TS would count as evidence for the neurodiversity paradigm? Limited representation of TS in Disability Pride may seem to undermine the neurodiversity paradigm. At the same time, given both Disability Pride's emphasis on physical disabilities and the social stigma associated with TS,<sup>25,26,29</sup> there may be barriers to TS representation in Disability Pride that have little to do with whether TS is pathologic.

Although both pragmatic and theoretical questions remain, existing tools from sociology and demography should be adequate to scientifically investigate membership in Disability Pride, rendering the central claims of the neurodiversity paradigm open to empirical investigation.

## Unique Strengths

Some have argued that many neurologic disorders come with relative strengths that outweigh, or balance out, associated deficits.<sup>30</sup> Paul Dirac, the Nobel Prize winning physicist, may be someone whose condition (possible autism) underpinned their concretism, scientific exactitude, and thus, success.<sup>31</sup> Relatedly, TS may be associated with verbal strengths in children; there may be a connection between TS and creativity; and children with TS may demonstrate higher cognitive control compared with neurotypical peers.<sup>32-34</sup>

Investigation of strengths associated with TS offers a third route to operationalizing the claims of the neurodiversity paradigm because they apply to TS. As before, challenges remain. First, comparative strengths and deficits will vary from person-to-person, making it difficult to draw general conclusions regarding the balance of the 2 across patients with TS. Second, there is limited exploration of the strengths associated with TS; extant research largely focuses on classifying and addressing deficits. Thinking about neurologic disorders in strengths, and not merely deficits, will require a substantial conceptual shift. Third, physiologic and cognitive measures of strengths and deficits are not straightforwardly related to everyday function.<sup>35</sup> Comparing various peaks and valleys of uneven physiologic or cognitive profiles is not sufficient to provide a fulsome view of functional strengths and deficits. Additional steps, likely including observation of patients in real world contexts, will need to be taken to consider how those peaks and valleys affect daily life. Nonetheless, investigation of the strengths associated with TS is empirically tractable and offers a third way to scientifically investigate the claims of the neurodiversity paradigm because they relate to TS.

## Suggestions and Future Directions

Although the neurodiversity paradigm's claims remain controversial, they are also gaining traction with academic and lay audiences. Neurologists should increasingly expect to encounter patients sympathetic to the neurodiversity paradigm and with associated antitherapy sentiments. At the same time, the serious challenges associated with living with TS, and the antitherapy arguments of some neurodiversity proponents, present a dilemma for the ethical conduct of both neurologic research and practice. Is it ethical to try and *treat* or *cure* TS? How can we try and answer this question, and what steps should be taken in the meantime?

## From Philosophical Idea to Empirical Construct

The neurodiversity paradigm challenges core assumptions and stigmas surrounding neurologic disorders and provides a conceptual foundation for political activism.<sup>36</sup> The neurodiversity paradigm was, however, never intended as a tool for use by neurologists. Nonetheless, neurologists need a way of assessing the truth of the neurodiversity paradigm's claims because they apply both to TS and to the full range of neurologic disorders.

Although genuine challenges remain, the core claims of the neurodiversity paradigm are empirically tractable. We have highlighted 3 potential routes to operationalization that would allow for the neurodiversity paradigm to shift from a philosophical idea and political tool to an empirical construct. Operationalizing the central claims of the neurodiversity paradigm is a key first step in understanding the implications of the neurodiversity paradigm for neurologic research and practice. It is likely also an important step in avoiding the conflict and crisis that has come to characterize the relationship between other health care disciplines that intersect with the claims of the neurodiversity paradigm and the patient populations they aim to help.

## Recognizing the Role of Neurology in Shaping Societal Views on Neurologic Disorders

Medical institutions play a central role in shaping society's understanding of disability, and neurology likely plays a similarly central role in shaping society's understanding of TS and other neurologic disorders.<sup>37</sup> Neurology is thus uniquely positioned to help alleviate the stigma faced by patients with TS. By shifting to an approach that views TS as a source of strengths, and not merely of deficits, neurology may help alleviate social stigma and provide society with a more accurate understanding of TS.

Whether, at a theoretical level, TS is best understood as a pathology, there is growing evidence that patients with TS often have relative strengths compared with their neurotypical peers.<sup>32-34</sup> By embracing an individualized understanding of TS and similar neurologic disorders that encompasses both unique challenges and patient profiles as they play out in the lives of particular patients, neurologists can not only help



destigmatize neurologic disorders but also advance our scientific understanding of the multifaceted nature of brain differences.

## Recognizing the Role of Neurology in Shaping Patient Views on Neurologic Disorders

Just as neurologists likely play a key role in shaping societal understanding of neurologic disorders, they can similarly shape how individual patients understand their conditions. A diagnosis of TS may be a patient's first introduction to the condition. Neurologists should thus be cognizant of the way in which their discussion of TS, and other neurologic disorders, can shape a patient's self-understanding. Following a diagnostic discussion, a patient with TS may leave the clinic on trajectory for the self-stigma too often seen in patients with TS.<sup>38</sup> Alternatively, given a different diagnostic conversation, a newly diagnosed patient may leave the clinic understanding that they have a "disease of the onlooker"<sup>21</sup>: a condition that comes with unique challenges, with unique strengths, and a condition that is a source of pride for many.

## Listening to Patient Voices and Precision Neurology

Neurologists can increasingly expect to encounter patients sympathetic to the neurodiversity paradigm and concomitantly resistant to therapy. Nonetheless, the challenges associated with neurologic disorders remain; neurologists often offer patients their best hope of getting needed help. How can neurologists square patients' needs with the antitherapy views that some patients may have?

A disconnect between patient and medical priorities is at the core of the tension between the neurodiversity movement and institutional medicine. Although neurodiversity activists push for greater focus on accessibility and assistive technology, medical practice and research often focus on treatment and cure. Centering patient voices in both clinical and research contexts is thus likely to be essential to successfully navigating neurology in the era of neurodiversity.

Patient-centered care has a long history in neurologic practice; the skills used in shared decision-making remain an invaluable vehicle for making patient values central to care.<sup>39</sup> Neurologists should recognize that patient priorities may emphasize accessibility and place little value on either treatment or cure; understanding that patients may reject pathologizing views of neurologic disorders may become an increasingly important starting point in neurologist-patient interactions. Ideally, neurologists should be able to pivot nimbly between thinking in the treatment of pathology, and the fiduciary responsibility to offer the most humane support for patients who may eschew or fear a *treatment* or *cure* for their condition.

Emerging technology, especially artificial intelligence (AI), may allow neurologists to buttress their practice of patient-centered care with a type of precision neurology not

previously possible. Machine learning has already afforded a level of fine-grained analysis that puts pressure on traditional TS diagnostic criteria.<sup>40</sup> Machine-learning technology may allow individualized analysis of the connections that hold between brain, behavior, environment, and function in a way that affords neurologic intervention in exactly those contexts in which TS presents as an obstacle for a particular patient. AI may thus provide an invaluable complement to patient-centered care, allowing neurologists to realize interventions that are responsive to patient values at a level of precision and granularity not previously available.

Centering patient voices in research contexts is, comparatively, more difficult. Discovering the research-related priorities of patients with TS will require empirical investigation. It is unlikely that attitudes toward research priorities are univocal. Nonetheless, research that is not responsive to the priorities of the patient population it aims to benefit is, minimally, unlikely to deliver the desired return on investment. More drastically, the ongoing pause of Spectrum 10k illustrates the cost of failing to adequately take into account the priorities of potential research participants.

## Conclusion

Neurology plays a central role in shaping both society's understanding of TS and the self-understanding of patients with TS. An exclusive focus on the deficits associated with TS and related neurologic disorders thus likely contributes to the stigma—both internal and external—with which patients with TS struggle. By shifting from a deficit-centric understanding to a more nuanced approach whereby neurologic disorders are understood in both strengths and deficits, neurologists can work to ameliorate the ableist bias with which patients with TS are confronted while simultaneously building a more fulsome understanding of the nature of neurologic differences.

Beyond highlighting the harms of ableist stigma, the neurodiversity paradigm challenges core assumptions about the correct way to understand the nature of neurologic disorders. Initially developed as a political tool and as a neurophilosophical construct, the central claims of the neurodiversity paradigm are nonetheless empirically tractable. Although obstacles to operationalization remain, work on scientifically investigating the central claims of the neurodiversity paradigm should commence forthwith.

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