Oh, Georges! What Have They Done to Your Beautiful Name?

“Georges Albert Édouard Brutus Gilles de la Tourette was a French physician and the namesake of Tourette’s syndrome, a neurological condition characterized by physical and verbal tics.”

The above is quoted from Wikipedia (accessed May 29, 2021). Indeed, the common name of the disease in the scholarly English literature, especially American, is “Tourette syndrome.” However, this is a misnomer. The family name of the neurologist who described this illness is Gilles de la Tourette, not just Tourette (Fig. 1).

Charles Edouard Brown-Séquard was a Mauritian neurologist who described what is now called Brown-Séquard syndrome. Nobody calls it the Séquard syndrome. Félix Vicq-d'Azyr characterized a nerve pathway in the brain named after him: It is the fasciculus Vicq d'Azyr (the mammillothalamic tract). Nobody calls it fasciculus d'Azyr. Guillaume-Benjamin Armand Duchenne de Boulogne who lived in the 19th century is labeled “the father of electrotherapy.” His family name is Duchenne de Boulogne. A prominent US neurologist working at Stanford University and one of the pioneers of closed-loop deep brain stimulation (DBS) is Helen Brönte-Stewart.1 Nobody calls her Stewart.

Concerning Georges Gilles de la Tourette, it is a rule without exception that in the French scholarly literature, the syndrome is always called “syndrome de Gilles de la Tourette.” Even in the British scholarly literature, eminent British neurologists and neuropsychiatrists always display the full name of the syndrome in the title of their publications.2-5

When Gilles de la Tourette published his seminal work, he titled it “la maladie des tics.” His mentor, Charcot, gave this disease the name “syndrome de Gilles de la Tourette” in honor of his student, adding, “Quel joli nom pour une maladie si horrible” (“What a beautiful name for so horrible a disease”).

The very first paper on DBS as a treatment for this disease, published by Vandewalle et al in The Lancet in 1999, was titled “Stereotactic treatment of Gilles de la Tourette syndrome by high frequency stimulation of thalamus.”6 Vandewalle stated in her article that she was inspired to perform DBS based on an old publication by Hassler et al on ablative stereotactic surgery for that condition; the title of that article, the Hassler paper that inspired Vandewalle, was “Stereotaxic treatment of tics and inarticulate cries or coprolalia considered as motor obsessional phenomena in Gilles de la Tourette’s disease.”7

So, how come the family name of the man and the name of the illness that he described have been amputated? It is a matter of respect to the name and legacy of that pioneer neurologist, and to his mentor, that we should strive to reinstate the real name of the illness as “Gilles de la Tourette syndrome” in all related publications.

Although we admit that the full name of Georges Albert Édouard Brutus Gilles de la Tourette is certainly too long and should be simplified, let us nonetheless abide by a quote of Albert Einstein: “Alles sollte so einfach wie möglich gemacht werden, aber nicht einfacher” (“Everything should be made as simple as possible, but not simpler”).

Statement of Ethics

We confirm that we have read the journal’s position on issues involved in ethical publication and affirm that this work is consistent with those guidelines. The present study is a Viewpoint about the name of Gilles de la Tourette and does

not include study or research on patients or animals and therefore does not need evaluation by an ethical committee.

Data Availability Statement
No data available/not applicable for this submission.

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References

Copy Number Variation in Parkinson’s Disease: An Update from Sub-Saharan Africa

Genetic mutations, including copy number variations (CNVs), in well-established Parkinson’s disease (PD) genes, have been identified as causes of familial and sporadic PD.1 These CNVs have primarily been detected in individuals of Asian and European ancestry1,2 but have rarely been reported in individuals of African ancestry from Sub-Saharan Africa (SSA) (Table 1).3,5,8,10 This may in part be due to the scarcity of SSA studies screening for CNVs, with only 96 unrelated PD cases having been screened to date (Table 1).3,10 To help address this critical knowledge gap, we used the multiplex ligation-dependent probe amplification (MLPA) assay to investigate CNVs within known PD and parkinsonism genes in 131 PD cases of African ancestry from South Africa and Nigeria.

Using MLPA kits (P051 and P052) that additionally detect the presence of the LRRK2 Gly2019Ser mutation, we screened 61 Nigerian and 70 South African PD cases for CNVs as described in Appendix S1. Neither the most common LRRK2 mutation nor known CNVs were detected. However, we acknowledge that CNVs in genomic regions not included in the MLPA kits may have been missed in this study. Initially, false-positive deletions were noted in two individuals (Appendix S1) as a result of two single-nucleotide variants (rs566749983:C>T and rs226249:T>C) that are reported to be more common in African than in European genomes.11 These false-positive findings not only highlight the importance of confirmation of MLPA results using a different technique in genetically diverse and understudied populations but also highlight the need for considering ancestry-specific markers in the design of genomic assays.

Most notably, we show that the genetic basis of PD in SSA is still severely understudied, given that with only 131 individuals, this is the largest SSA study to date screening African ancestry PD cases for CNVs. Combining our results with previous studies on African ancestry cases in SSA (Table 1), the frequency of CNV mutations in PRK2 is 2.2% (5/227 unrelated cases) but remains 0% in other PD genes, including SNCA and PINK1. These findings (together with the fact that LRRK2 Gly2019Ser, one of the most common PD-associated mutations,12 was not present in our study participants, and that it remains undetected in SSA individuals of African ancestry)1,13,14 support the notion that PD genetics is understudied in SSA and reinforces that the genetic contributors to PD in SSA may differ from those identified in other ancestries. Therefore, although challenging,15 further research into the genetic causes, including CNVs, of PD in SSA is warranted. For this, we promote across-border collaborative research between clinicians and researchers (as illustrated in our study) to achieve greater sample sizes, and we promote the use of next-generation sequencing or long-read sequencing of whole nuclear and mitochondrial genomes.

Ultimately, the inclusion of diverse populations in future PD studies, as highlighted in the Aligning Science Across Parkinson’s/Global Parkinson’s Genetics Program project (ASAP/GP2; https://parkinsonsroadmap.org/) and supporting consortia efforts (e.g., Genetic Epidemiology of Parkinson’s Disease [GEoPD] and International Parkinson’s Disease Genomics Consortium [IPDGC]), will be key to identifying novel genetic contributors to disease and gaining a better understanding of PD’s complex etiology on a global scale.

Statement of Ethics
Ethics approval for this study was obtained from the Health Research Ethics Committee at Stellenbosch University (Protocol numbers: 2002/C059, S20/01/010, and N16/04/041).