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Perspective

Striving for a Realistic and Unapologetic View of Huntington's Disease

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Abstract. This article describes how the author, a Huntington's disease (HD) gene expansion carrier and long-time advocate, has helped give voice to the HD community through his blog, *At Risk for Huntington's Disease*. Since 2005, the 321 articles have helped document the new and harrowing experience of living in the gray zone between a genetic test result and disease onset. *At Risk for HD* has explored major challenges for the HD community, has become a key reference for HD families, and has chronicled the quest to defeat the disease. This article analyzes the blog's thematic impact.

Keywords: Huntington's disease, Gene Veritas, blog, genetic testing, science reporting, therapies, stigma, support network, patient advocacy

Those facing the devastating disorder of Huntington's disease (HD) must grapple with enormous challenges and commitments. Affected individuals and their families have reacted in multiple ways. This author, a long-time HD advocate, has used his background as a historian and writer to give voice to the HD community through the blog *At Risk for Huntington's Disease* (https://www.CureHD.blogspot.com). The blog describes my personal confrontation with the disease, its overwhelming impact on families, and the hunger for therapies, serving as a chronicle of the quest to defeat the disease. This article (re)introduces *At Risk for HD* as a key resource for researchers and HD families, analyzes its thematic impact, and, thankfully, marks the extraordinary feat of an HD gene expansion carrier continuing to blog in his 60 s.

In January 2005, aiming to cope with and safely share my fears as a person with 40 CAG repeats, I started *At Risk for HD* under the pseudonym "Gene Veritas," the "truth in my genes." I hoped to generate greater awareness about this rare disease and encourage donations for therapeutic research. As I unburdened myself, the blog helped me build a support network, as others writing about devastating diseases have done [1], and led me to better understand the many dimensions of HD. In the ensuing 18 years, the blog's 321 articles have helped document the harrowing, new experience of living in the gray zone between a genetic test result and disease onset. *At Risk for HD* has explored HD's debilitating symptoms and psychosocial ramifications. To quote a fellow gene expansion carrier regarding my portrayal of families facing wrenching decisions about family planning and abortion, the blog has exposed HD in a "realistic and unapologetic way" [2]. To foster therapeutic progress, I have strived both to translate key scientific developments into nontechnical language and to encourage participation in research studies and clinical trials. In 2012, I took a definitive step out of the "HD closet" by using my real name in a *Chronicle of Higher Education* essay [3], but "Gene Veritas" lives on as a trademark of my advocacy. It symbolizes the public-versus-private dilemma stoked by the deep stigma and denial associated with HD. Despite

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the inevitability of my onset—new research better defines the stages of the disease, in which progression begins at birth [4]—I have preferred to keep the ambiguous "at risk" in my blog title. I continue to refer to myself as an HD gene expansion carrier, but not as "a person with HD," a term ever more present in the scientific literature [5]. With the important but complex scientific terminology of HD I continually face the challenge of explaining the seriousness of the disease and its implications [6], including the risk of affected individuals internalizing stigma [7, 8]. Also, whereas my mother developed symptoms in her late 40 s, I have reached my early 60 s without a diagnosis of onset. My hope for a very late onset forms part of my strategy of maintaining a positive attitude.

Table 1 indicates the number of times a particular topic was discussed in the 314 blog articles produced by April 2022; many articles addressed multiple topics. The most frequent topic was advocacy and awareness-building. Close behind was scientific research, followed by clinical trials. The table demonstrates HD's many repercussions for affected individuals and their families. Two topics very significant for expansion carriers and untested at-risk individuals stand out: coping strategies before onset, and fear of onset and symptoms/living at risk. Many articles referred to family members, as well as my own family's struggles: after a two-decade fight, my mother's death from HD in 2006 at the age of 68, my test for the expansion in 1999, and my daughter's negative prenatal test in 2000. Such bioethical concerns were raised regularly, including exiting the HD closet, genetic testing, stigma and discrimination, and preimplantation genetic diagnosis. *At Risk for HD* has also emphasized caregiving. I have explored how religion, faith, and spirituality provide meaning and purpose for myself and others, including ample coverage of Pope Francis' historic 2017 audience with the HD community.

Table 1

Topic	Times discussed
Advocacy, awareness-building	139
Scientific research	122
Clinical trials	84
Solidarity	74
Coping strategies before onset	68
Information about HD, the affected	63
Fear of onset, symptoms/living at risk	62
Waiting for a cure	46
Mother/grandmother	44
Participation in research, clinical trials	40
Exiting the HD closet	36
Genetic testing	33
Stigma, discrimination	33
Spouse	31
Daughter/son	29
Caregiving	28
Preimplantation genetic diagnosis and prenatal testing	28
Brazil, Latin America	27
Religion, faith, and spirituality	26
Juvenile HD	16
Coping strategies after onset	15
Bioethics	14
Denial	13
Relinquishing personal, professional dreams	12
Nursing home care	12
HD in the media	12
Mortality	11
Relationship with physicians, researchers	8
Brain trauma in athletes	7
Suicide	6
Family tensions, conflict	5
Science-faith connection	5
Financial impact	5
Abortion	4
Preciousness of time, life	3
End-of-life care	2
Courtship, relationships, marriage, and love	2
Psychotherapy	2
Open science	2
Medical costs	1
Adoption	1
Care versus cure	1

At Risk for HD has captured major challenges facing the HD community. In 2011, I contrasted two families' difficult decisions regarding abortion: one couple opted to abort an expansion-carrying fetus [9], while a juvenile-HD-afflicted woman chose to have her untested baby [10]. A 2014 article described a video of police mistreatment of a man suffering from HD—"I can't breathe!"— and revealed how rare disease communities must fight against ignorance, discrimination, and hostile treatment [11]. I reported in depth on the critical 2015 meeting of HD advocates and family members with the U.S. Food and Drug Administration regarding patient-focused drug development [12]. Since 2008, the blog has tracked the efforts by Ionis Pharmaceuticals, CHDI Foundation, and Roche to pioneer a therapy using an antisense oligonucleotide to lower the mutant huntingtin protein in the brain. Started in 2011 and focusing solely on developments in research, the online portal HDBuzz.net has crucially contextualized this information, and combatted misinformation, in articles authored by scientists [13, 14]. *At Risk for HD* offers the viewpoint of a *non*-scientist HD family member. The blog has frequently featured interviews, including many on video, with leading scientists as well as activists. Informed by the trailblazing work of advocate-historian Alice Wexler [15–17], *At Risk for*

HD has also sought to cast the HD cause in historical perspective, recognizing the context of other rare and neurodegenerative diseases.

At Risk for HD has put into words feelings and experiences that some may not verbalize or share because of the stigma, shame, embarrassment, and denial connected with HD. In some instances, as both publicly posted and private comments confirm, the blog has enabled people to better cope, making their HD journey less solitary. For this author, reporting on the quest for therapies and building ties to scientists, clinicians, advocates, and affected families has functioned not just as a personal coping mechanism, but a service and source of comfort for a community consumed by urgency. With 1.47 million hits since its inception [18] and a global following through Facebook, HD news networks and e-mail, the blog has reached readers from grassroots activists to biopharma CEOs. Though immersed in the fight, I pursue objectivity and accuracy through good journalistic practices, scholarly rigor, and regular feedback. Readers have offered countless expressions of support, as well as constructive criticism. I yearn to post an article about the approval of a disease-modifying treatment—and the end of fear of Huntington's. From researchers to local volunteers, we can all contribute to this cause with our unique talents.

Kenneth P. Serbin (aka Gene Veritas) is a professor of history at the University of San Diego. His research spans the fields of Brazilian social, cultural, political, and religious history, as well as the history of science, technology, and medicine. In 2011 he was named the Person of the Year of the Huntington's Disease Society of America. He is currently investigating the social and scientific history of the HD movement.

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CONFLICT OF INTEREST

In solidarity with the HD cause, Kenneth P. Serbin holds a symbolic amount of Ionis shares.

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