

International opinion survey on nomenclature for platelet-type von Willebrand disease: communication from the SSC of the ISTH

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Abstract

Background: The nomenclature for platelet-type von Willebrand disease (PT-VWD) remains an open debate, with several proposed names in use in the literature for this rare inherited platelet disorder. It has been suggested that this name may lead to delays in diagnosis since the diagnostic pathway of patients often starts with von Willebrand factor assays, while the disorder is due to pathogenic gain-of-function (GOF) variants in the platelet *GP1BA*.

Objectives: This joint International Society on Thrombosis and Haemostasis (ISTH) Subcommittees' project aimed to engage with the international community on the disease's nomenclature.

Methods: A working group, established under the auspices of 3 Subcommittees, explored issues around nomenclature and proposed possible names with input from a

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panel of 6 global experts. Two rounds of surveys were administered online via ISTH REDCap and My ISTH Community.

Results: The 4 names, GPIBA GOF disorder, PT-VWD, glycoprotein Ib α GOF disorder, and PT-pseudo VWD, were selected in the first survey round by 40% to 60% of participants. The first 2 were the top choices and showed almost equal support (28% and 24%, respectively). A second focused round did not generate consensus for name change.

Conclusion: Based on the survey results, the decision was in support of retention of the more commonly used nomenclature (PT-VWD). More work is needed to ensure this name is adopted universally and in all current online databases to achieve consistency.

KEYWORDS

blood platelet disorders, platelet aggregation, von Willebrand Disease, platelet type, ristocetin, rare disease

1 | INTRODUCTION

Platelet-type von Willebrand disease (PT-VWD) is an autosomal dominant bleeding disorder resulting from gain-of-function (GOF) mutations in the platelet GP1BA gene, causing an enhanced affinity of binding von Willebrand factor (VWF) by platelets, resulting in the clearance of hemostatically active VWF multimers and platelets, with subsequent mucocutaneous bleeding. Despite the typically encountered mild to moderate bleeding tendency, life-threatening bleeding can occur in thermostatically challenging conditions such as infection, pregnancy, and surgery [1]. PT-VWD remains a frequently underdiagnosed and misdiagnosed disorder as it is often confused with type 2B von Willebrand disease (VWD), which displays a similar phenotype as PT-VWD but is caused by mutations in the A1 domain of VWF gene, leading to enhanced VWF binding to platelets, rather than in the GP1BA gene. VWD is believed to be the most common bleeding disorder, affecting approximately 1% of the population, with type 2B VWD making up 5% to 10% of these diagnoses. Approximately 15% of patients diagnosed with type 2B VWD display GP1BA mutations consistent with PT-VWD [2]. The disease can also be misdiagnosed with other conditions of unexplained thrombocytopenia or immune thrombocytopenic purpura. While the international PT-VWD registry (www.pt-vwd.com) reports only 55 patients, recent evidence from population-based genetic epidemiology analysis indicates PT-VWD is not as rare as previously thought and the disease indeed remains under or misdiagnosed [3].

Four members of 1 family with a mild bleeding disorder were reported in 1982 by Harvey Weiss et al. [4], who identified in platelets the basic defect leading to reduced high-molecular-weight VWF multimers in plasma and named this condition pseudo-VWD. Independently and in the same year, Miller and Castello described another family with the same phenotype and named it PT-VWD [5]. It is interesting to note that in 1981, a Japanese group led by Takahashi [6,7] already reported a similar phenotype and suggested the identification of a new disorder but without naming it. In addition to pseudo-VWD [4] and PT-VWD [5],

other names have been proposed over the years, including "VWD-mimic" disorder [8] and "platelet-type pseudo-VWD" [9].

Although the term PT-VWD is in common use, this nomenclature is potentially misleading. The diagnostic workup of patients with PT-VWD often starts with VWF assays, while the disorder is due to platelet GP1BA GOF variants and not VWF variants [10]. Both the diagnostic and therapeutic approaches to PT-VWD and VWD are quite different, and management appropriate for those with VWD can be detrimental to patients with PT-VWD or requires extra caution, eg, 1-deamino-8-D-arginine vasopressin [11]. For physicians less experienced in hemostasis, this subtlety may be lost, resulting in inappropriate use of VWD-directed therapies. The reconsideration of the nomenclature of this condition might help to delineate the disease as distinct from VWD, with specific diagnostic and management considerations.

Finally, the more common and widely diagnosed platelet function defect, Bernard-Soulier syndrome, results from loss-of-function variants in GP1BA; some of which are close to the amino acids described as being pathogenic for PT-VWD [12]. Thus, a disease name with information about the gene and the nature of the mutation (GOF) could be considered.

Based on all the above, a joint initiative of the Subcommittees (SSCs) on Platelet Physiology, Genomics in Thrombosis and Hemostasis (TH), and VWF was started to revisit the disease's nomenclature, considering the evolving scientific landscape of inherited platelet disorders [13,14], and in consultation with International Society on Thrombosis and Haemostasis (ISTH) members and the scientific community.

2 | METHODS

A small working group (M. Lavin, R.L., P.G., and M.O. [Chair]) was established to design a collaborative project under the auspices of 3 SSCs (Chairs: M. Lordkipanidzé, R.B., and K.F.) to examine and standardize the current nomenclature of PT-VWD. The working group met on June 2, 2021, to discuss the project action plan, issues around

nomenclature, and proposals for a new nomenclature. The group then nominated a panel of worldwide experts to review and discuss the newly proposed names and their rationale. The collaborative expert panel was formed by physicians and scientists who had studied, published, and/or cared for patients with this disease. Seventeen experts were contacted. The following 6 responded to the survey: Harvey Weiss (USA), Johnathan Miller (USA), Jose Lopez (USA), Analia Sánchez Luceros (Argentina), Gillian Lowe (UK), and Emmanuel Favaloro (Australia). Additionally, the following experts supported the survey design: Jecko Thachil (UK), Shirin Ravanbod (Iran), and Suchitra Acharya (USA).

A 4-question survey (Supplementary Table S1) was designed to capture the views of ISTH members and scientific community. The survey addressed whether a change in nomenclature is required, listed 8 names with rationale for each one, and requested participants to rank their top 3 choices with an opportunity to suggest new names. The last 2 questions asked about the ideal ISTH SSC to discuss issues related to this disease, and to which SSC the participant belonged. The survey and proposed nomenclature were reviewed and approved by 3 SSC chairs and the expert panel prior to distribution. The survey was administered online via ISTH Research Electronic Data Capture and posted on the ISTH Platelet Physiology SSC website between July 6 and August 30, 2021. Additionally, the survey was sent via email to the members of 3 SSCs. The survey was also publicized via a presentation by the SSC on Platelet Physiology (ISTH 2021 in Philadelphia/virtual meeting) to inform the community about the project, proposed nomenclature, and to encourage broad participation.

Criteria for nomenclature acceptance were set by the working group *a priori*. It was agreed that if needed, the top 2 results for naming the disorder from the first survey would form the basis of a second survey. Additionally, if new suggestions for a new name (not previously proposed/discussed by the group) were provided by 10 or more respondents, they would also be included in survey 2. Any new option should have reached 60% in survey 2 to allow acceptance. If the vote was <60%, a poll by SSC chairs and co-chairs (each member with 1 equal vote) would resolve this. If tied, final deciding vote would lie with the 3 SSC chairs.

3 | RESULTS AND DISCUSSION

3.1 | Survey 1

3.1.1 | Participant characterization and survey completion

Overall, 155 entries were recorded for the survey link. Participants were asked to indicate the SSCs they were currently involved with and were given the ability to indicate if they were part of multiple SSCs or SSCs other than the 3 co-leading SSCs of this project. Results showed that 74 participants indicated their involvement in the VWF SSC, 54 in the Platelet Physiology SSC, 28 in the Genomics in TH SSC, and lastly, 57 entries were recorded for "other ISTH SSCs." Therefore, the bulk of participants were members of VWF (74%), Platelet

Physiology (45%), and Genomics in TH (28%) SSCs. This spread of involvement across all ISTH SSCs can be seen in Figure 1. It is to be noted that some participants were members of >1 SSC.

Incomplete answers, characterized by failing to respond to all 4 questions, were 4.5% (7/155) of the total entries. However, all 155 participants answered the "NAMES" question, and thus 7 incomplete entries were not excluded from the analysis of the data. Around 10% (16/155) of the recorded entries were submitted by members of the collaborative expert panel.

3.1.2 | Do you support the need for change of current name? Respondents were first asked to indicate whether they supported the need for change of the most widely used current name, PT-VWD

Of 155 participants, 66% (103) answered yes, 31% (48) no, and 3% (4) did not answer. The 48 respondents still provided their top 3 choices for names that could replace PT-VWD and answered the remaining questions.

3.1.3 | Participant selections of top choices

Participants were provided with various options, together with a brief rationale for each option. They were asked to rank their choice of 1, 2, and 3 out of a list of 8 potential names. Of 155 participants (who at least provided their top 1 choice), 28% (43) chose "platelet GPIBA GOF disorder (GPIBA GOF disorder)" as their rank 1 choice, making it the most selected option and the most agreed-on name among respondents. PT-VWD was selected by 24% (37) of 155 participants, and "platelet-type pseudo-VWD" was selected by 13% (20) of participants. "Platelet glycoprotein Iba GOF disorder (GPIba GOF disorder)" was selected by 9% (14) of participants. The 4 names were selected by 40% to 60% of participants among their 3 top choices. However, GPIBA GOF disorder appeared dominant in first choices and the overall top choice as well, and was a clear favorite among those who felt a name change was needed. It is worth noting that even among those who felt a name change is desirable, around 17% selected PT-VWD as one of their top choices. No new names were proposed. The number and percentage of all 155 participants and their choices are highlighted in Figure 2.

We sought to examine the name ranking provided by only those who supported the change of current name (66%; 103). Once again, GPIBA GOF disorder was the most popular. It was selected as top choice by 40% of participants. Platelet-glycoprotein (GP)Iba VWF disorder was selected by 15% (15) of participants. Next was GPIba GOF disorder, selected by 13% (13). The number and percentage of those 103 participants and their choices are highlighted in Figure 3.

Lastly, the top choice selections for members of the expert panel were tabulated. Based on the complete data available for 6 experts, 50% (3) preferred the current name (PT-VWD) as their top selection, while the remaining half expressed dispersed top choices. Specifically, each of the following was selected as number 1 by one of the expert respondents:

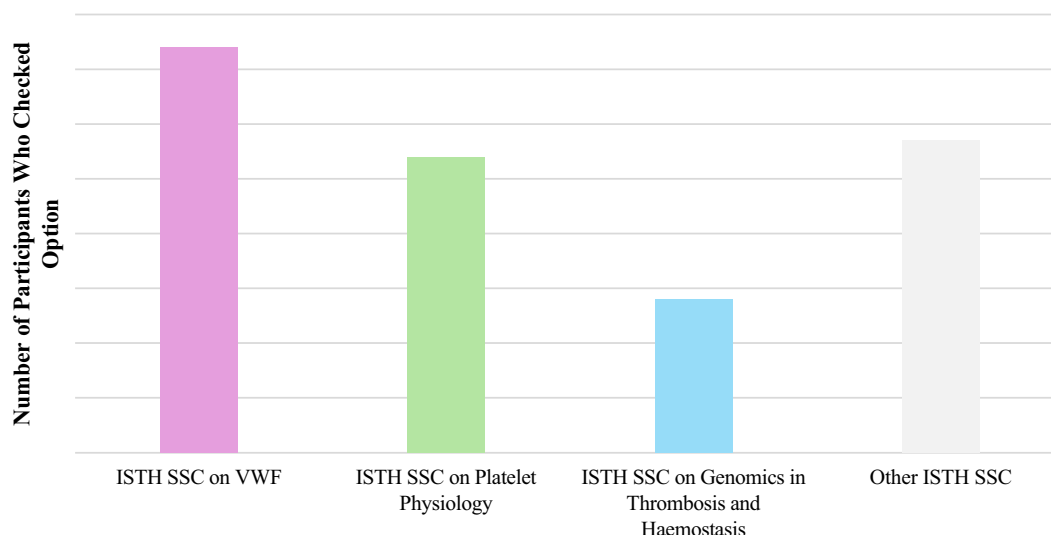


FIGURE 1 Subcommittee (SSC) membership of all 155 participants. Note that some members were involved in >1 SSC. ISTH, International Society on Thrombosis and Haemostasis; VWF, von Willebrand factor.

Bernard Soulier GOF, platelet-type pseudo-VWD, and pseudo-VWD. Overall, choices were highly variable, and none of the 6 expert members chose GP1BA GOF disorder as their first choice for a name.

3.1.4 | Where would it be most appropriate to discuss issues of this disease? When asked where issues related to this disease should be thoroughly discussed, ~59% of the 153 respondents who answered this question (91 entries) indicated this disease needed to be discussed in combination of all 3 SSCs (VWF, Platelet physiology, and Genomics in TH)

Thirty of 153 participants (20%) selected ISTH SSC on Platelet Physiology. Sixteen percent (24) and 5% (8) indicated that these issues should be addressed by the ISTH SSC on VWF and Genomics in TH, respectively.

3.2 | Survey 2

The data from the first survey indicated that the scientific community remained undecided regarding the most appropriate nomenclature. Thus, we proposed to reexamine the top 2 choices as shown in [Figure 2](#) (GPIBA GOF disorder; 28%) and the traditional nomenclature (PT-VWD; 24%) again in a second survey. This second round was comprised a single question ([Supplementary Table S2](#)). We asked participants to select their preference out of those 2 choices. Participants who were strongly opposed to either name were given an option to signal their disapproval. This survey was also administered online via ISTH REDcap, publicized via a presentation by the SSC on Platelet Physiology (ISTH 2022 in London, UK meeting), and posted on “My ISTH SSC community” platform for 1 month after the meeting. A

total of 136 participants completed the second survey, and the results showed 54% favored PT-VWD, whereas 46% favored GP1BA GOF disorder, and no one expressed strong opposition to either name ([Figure 4](#)).

3.3 | No consensus toward a change of PT-VWD nomenclature

Recognizing the top 2 names still ranked very closely following 2 survey rounds, we concluded that there is insufficient evidence to suggest a change in the nomenclature, leading to the retention of the traditional PT-VWD nomenclature.

3.4 | Summary and final remarks

The issue of nomenclature in PT-VWD has been discussed on several occasions in the literature [\[1,15,16\]](#), and in a 2007 review [\[17\]](#), it was stated that “until a final agreement on nomenclature is reached, we will continue to use the term platelet-type VWD, since this is the name that attained most popularity.” This current name, “PT-VWD,” has pros and cons. While it may accurately note that the basic defect is in platelets, it may also suggest the need to test both platelets and VWF. Moreover, “VWD” in the name may be confusing to the general hematological and medical community. Guidance on the diagnosis and management of PT-VWD by the SSC of the ISTH on Platelet Physiology has recently been reported, documenting the disease remains under or misdiagnosed worldwide [\[18\]](#). However, for physicians who are less experienced in hemostasis, the current nomenclature may still result in inappropriate use of VWD-directed therapies.

History and nomenclature of diseases represent important issues, particularly in rare diseases. Some guidance can be obtained from the

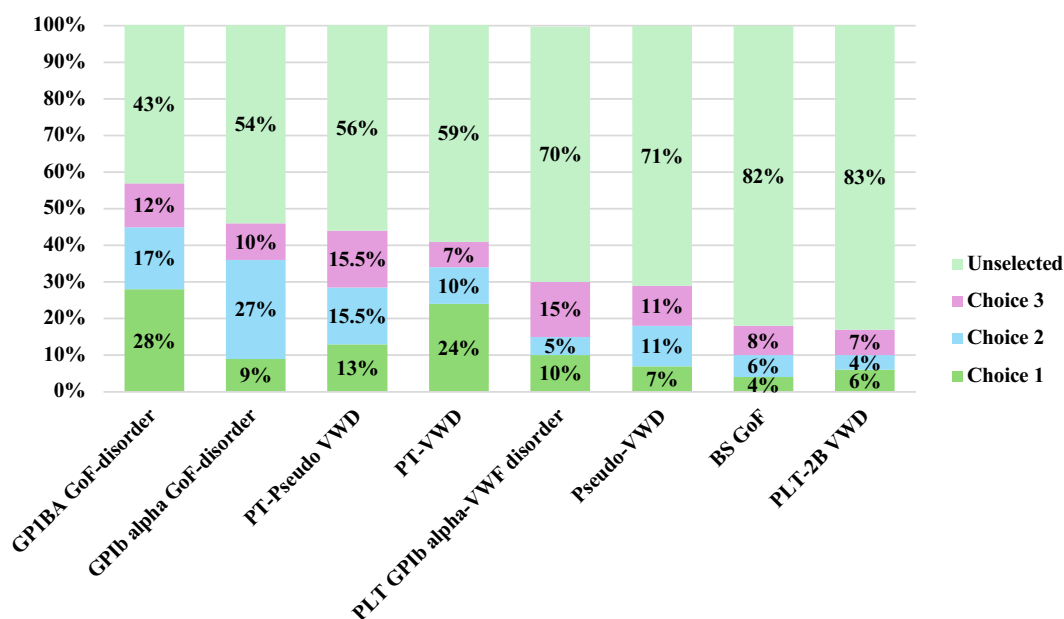


FIGURE 2 Survey 1 data. The number and percentage of all 155 participants and their top choices. Twenty-eight percent and 24% chose GPIBA gain-of-function (GOF) disorder and platelet-type von Willebrand disease (PT-VWD) as their top choice (choice 1), respectively. BS GOF, Bernard Soulier Gain-Of-Function; GPIb, glycoprotein Ib; PLT GPIb alpha-VWF, platelet glycoprotein Ib alpha-von Willebrand factor; PLT-2BVWD, platelet type-von Willebrand disease type 2B; Pseudo-VWD, pseudo-von Willebrand disease; PT, platelet type.

Orphanet database (www.orpha.net), which defines a set of rules to be used to promote a correct nomenclature. Nomenclature must, in general, be based on clinical practice and validated by experts in the

field; it should be self-sufficient and avoid ambiguity. Consistency and stability are important, but due consideration must be given to the evolution of scientific knowledge.

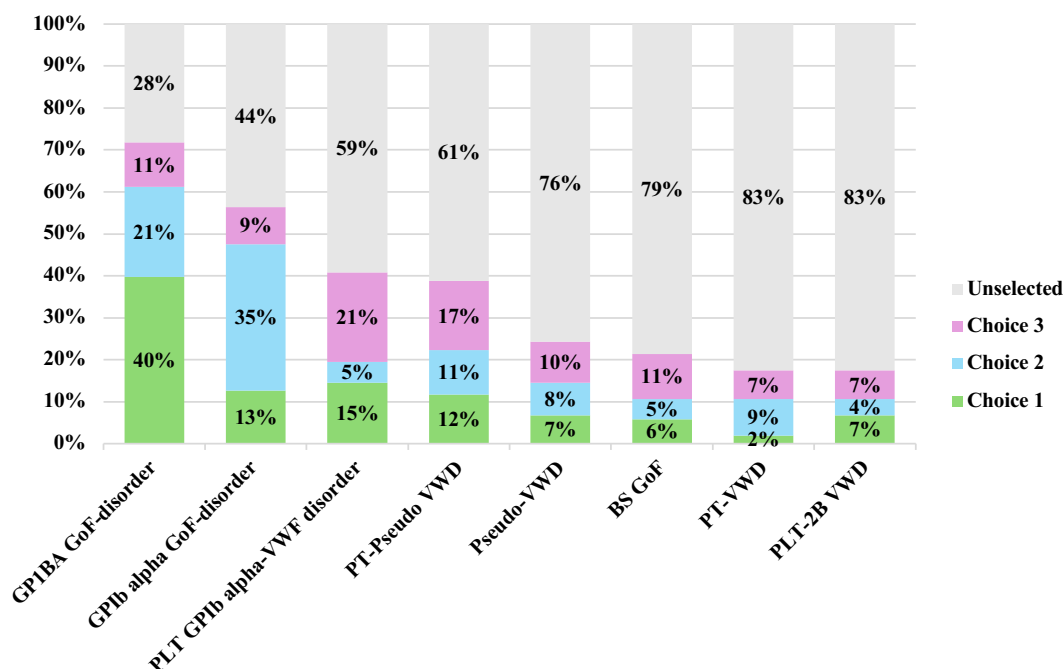


FIGURE 3 Survey 1 data. The number and percentage of the 103 participants who answered yes to change the name, with their choices highlighted. Forty percent of participants chose GPIBA gain-of-function (GOF) disorder as their top choice (choice 1). BS GOF, Bernard Soulier Gain-Of-Function; GPIb, glycoprotein Ib; PLT GPIb alpha-VWF, platelet glycoprotein Ib alpha -von Willebrand Factor; PLT-2BVWD, platelet type - von Willebrand Disease type 2B; Pseudo-VWD, pseudo-von Willebrand disease; PT-VWD, platelet-type von Willebrand disease.

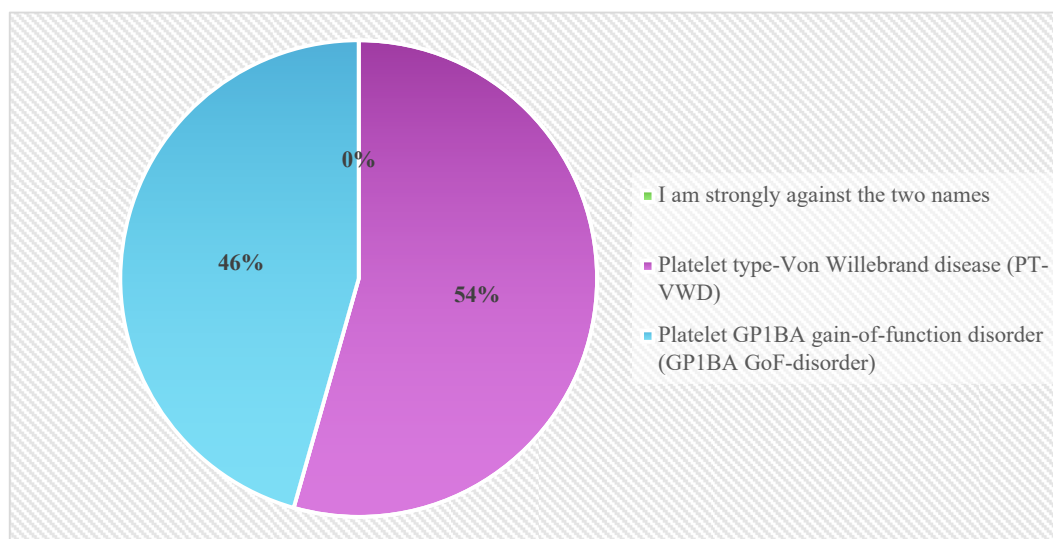


FIGURE 4 Survey 2 data indicating no consensus for nomenclature change. GPIBA GOF disorder, GPIBA gain-of-function disorder; PT-VWD, platelet-type von Willebrand disease

In this study, the first survey indicated a desire to change the current name, PT-VWD, to a new one that perhaps better represents the mechanism of the disease and potentially improves the directive actions for testing and diagnosis. However, the results of this survey showed there was no strong favoritism to any of the 8 proposed names, and the community remained undecided between “PT-VWD” and “GPIBA GOF disorder.” This may have been due to a larger shift in the commonly used nomenclature that perhaps the community was ready to endorse, but no other names were suggested in the surveys that rallied opinion toward a new nomenclature. The results of the second survey showed similar split in opinion; thus the decision was to retain the traditional name PT-VWD.

We have consulted with the genomics leaders at ClinVar, who indicated that the disease database Online Mendelian Inheritance in Man (<https://www.omim.org/>) still reports both names for this rare platelet disease: PT-VWD and pseudo-VWD (accession number: 177820). The Mondo Disease Ontology database (<https://www.ebi.ac.uk/ols4/ontologies/mondo>) uses pseudo-VWD (accession number: 0008332). Finally, the Orphanet Rare Disease Ontology database (<https://www.orpha.net/>) also uses pseudo-VWD (accession number: 52530).

It is worth mentioning that first confirmatory evidence that this is a genetic platelet bleeding defect was given in 1991 when a GOF variant was identified within the VWF-binding region of the platelet GPIBA gene [19], 4 years after the cloning of that gene by Lopez et al. [20] in 1987. GPIb α is the major subunit of the platelet GPIb-IX-V complex [21]. The ISTH PT-VWD Registry (www.pt-vwd.com) has listed 5 single-nucleotide variants within the VWF-binding domain of GPIb α and one 9-residue deletion further down in the mechanosensory domain, which is likely involved in the pathology of the disease [22]. Two other variants outside the VWF-binding site but still within the leucine-rich repeats domain of GPIb α have been recently described and functionally characterized [23,24]. Thus

far, a total of 9 GOF variants have been described in this disease [25].

Overall, a membership of 3 ISTH SSCs, together with a group of experts, 2 rounds of surveys, and a wide consultative process, highlighted a more or less even split in opinion toward retaining the traditional PT-VWD name and adopting a new nomenclature. When the community was asked to choose between the 2 top contenders, the survey rendered a similar result. Taken together, we have reached the conclusion that there is no sufficient scientific or cultural imperative toward recommending a nomenclature change from PT-VWD. Future efforts will focus on adopting this nomenclature universally across clinical databases while discouraging the use of alternative terminologies.

APPENDIX

EXPERT collaborators' contribution (survey construction and/or responses)

Harvey Weiss (NY, USA), Johnathan Miller (Chicago, IL, USA), Hoyu Takahashi (Tokyo, Japan), Jose Lopez (Seattle, WA, USA), François Lanza (Strasbourg, France), Jerry Ware (Little Rock, AR, USA), Paquita Nurden (Pessac, France), Analía Sánchez Luceros (Buenos Aires, Argentina), Suchitra Acharya (NY, USA), Shirin Ravanbod (Tehran, Iran), Will Lester (Birmingham, UK), Gillian Lowe (Birmingham, UK), Jecko Thachil (Manchester, UK), Said Enayat (Tehran, Iran), Emmanuel Favaloro (Sydney, Australia) are collaborators who have contributed to the design the study and questionnaire and/or voted for the choice of disease name.

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guidance [12] and the controversial issue of platelet-type von Willebrand disease nomenclature.

AUTHOR CONTRIBUTIONS

This is a joint project of 3 Subcommittees of the International Society on Thrombosis and Haemostasis: Platelet Physiology, VWF, and Genomics in Thrombosis and Haemostasis. A working group formed by M.O., M. Lavin, R.L., and P.G. developed the concept, proposed new names, and created the survey. R.B., M. Lordkipianidzé, K.F., and P.G. facilitated the study, led the international consultation process, and reviewed the study plan, surveys, and manuscript. EXPERT collaborators reviewed the study design and participated in surveys. A.E. administered the survey in REDCap and gathered and analyzed data. M.O. and A.E. analyzed data and wrote the first draft. All authors critically reviewed and approved the manuscript prior to submission.

DECLARATION OF COMPETING INTERESTS

M. Lavin has served as consultant for Sobi and CSL Behring, received speaking fees from Pfizer, and received indirect funding from Takeda for development of educational materials. M. Lordkipianidzé is a Canada Research Chair in Platelets as biomarkers and vectors. All other authors have no conflict of interest.

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SUPPLEMENTARY MATERIAL

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