

# Defining indications for thrombophilia testing – A modified Delphi consensus study in Switzerland

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Introduction

Thrombophilia is a hereditary or acquired condition associated with an increased risk of venous thromboembolic events (VTE) and VTE-recurrence. The clinical relevance of thrombophilia testing remains uncertain, and existing recommendations regarding patient selection for testing are inconsistent. This has resulted in considerable variability of thrombophilia testing in clinical practice nationally and internationally. This Delphi survey was conducted to establish expert consensus on thrombophilia testing in Switzerland.

Methods

A modified Delphi study was performed with clinical experts managing VTE-patients in Switzerland. A steering committee developed clinical scenarios and statements on thrombophilia testing. These were distributed to the expert panel who rated their agreement and provided written feedback. Consensus was defined as ≥70% of experts rating a statement ≥5 on a 7-point Likert scale.

Conclusion

In this modified Delphi process, an expert panel achieved consensus on 32 practical recommendation statements defining indications for thrombophilia testing. The consensus statements align with most existing recommendations, though some diverge (e.g., thrombophilia testing in unprovoked VTE). The consensus provides clear guidance for physicians on thrombophilia testing in specific clinical scenarios. Dissemination of this study to front-line clinicians will contribute to harmonised testing in Switzerland. By standardising strategies, these statements may prevent unnecessary testing, reduce variability in care, support consistent clinical decision-making, and ultimately improve patient counselling, management, outcomes and healthcare resource utilisation.

## Results

The survey was completed by 42 clinical experts. Consensus was reached after two rounds on 32 statements covering indications such as unprovoked VTE, provoked VTE (incl. hormonal risk factors), unusual site VTE, paediatric VTE, and patients without VTE. [Figure 1](#) shows an overview of the modified Delphi process.

**Strong consensus** supported hereditary and acquired thrombophilia testing in patients < 60 y.o. with unprovoked VTE or unusual-site VTE and in women with hormone-associated VTE.

**Consensus** discouraged testing patients with VTE and major transient risk factors and in asymptomatic individuals with VTE in family history where familial thrombophilia is unknown.

**No consensus** was reached on testing patients <60 y.o. with unprovoked VTE and an additional persistent VTE-risk factor.

[Figure 2](#) shows the practical consensus statements defining indications for thrombophilia testing.

Figure 1:

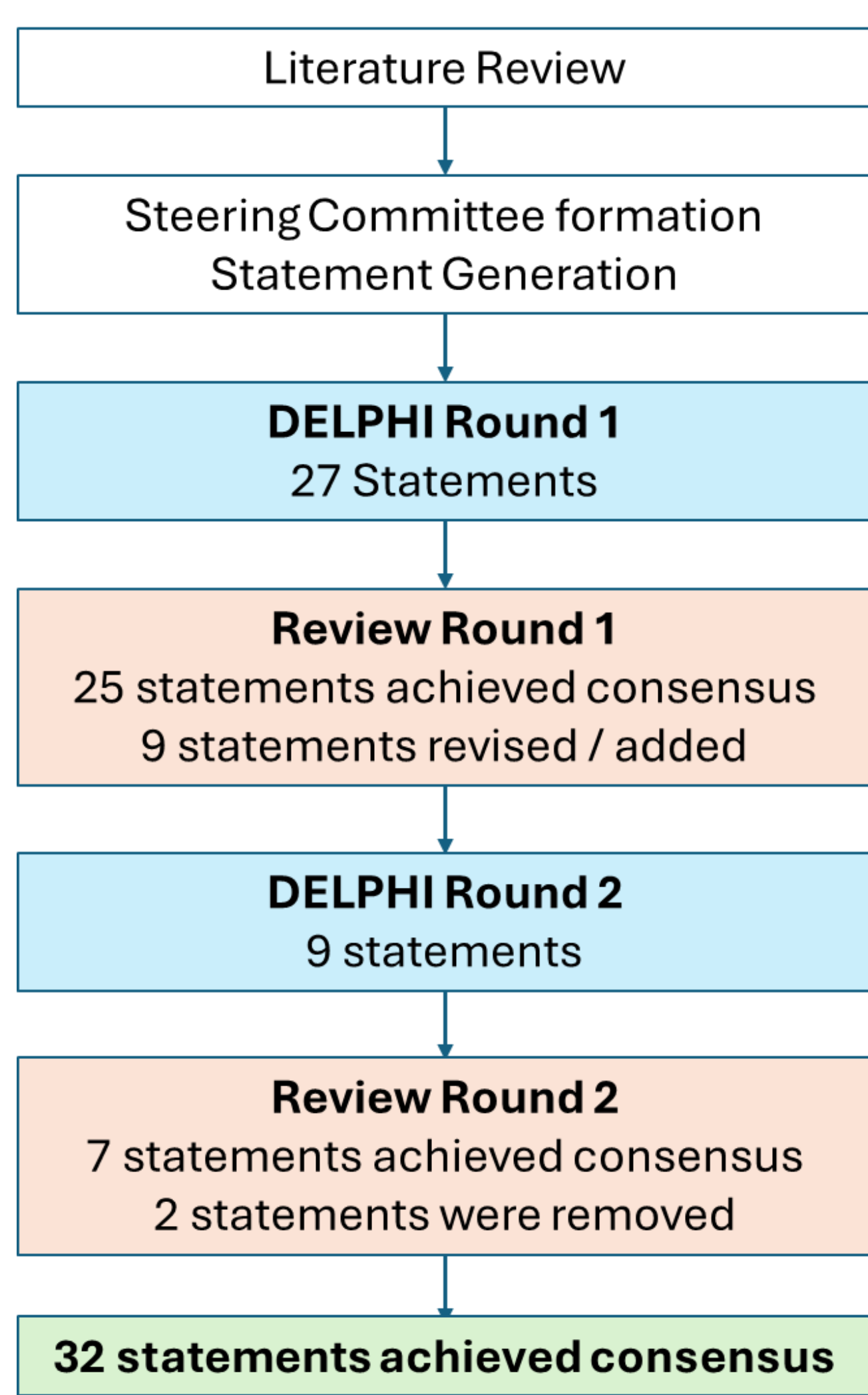


Figure 2:

Unprovoked VTE	Provoked VTE	No VTE in patient	No VTE in patient – women, hormonal	Paediatric VTE
<div>No clinical RF</div> <div>≤60 y.o. &gt;60 y.o.</div> <div>YES hereditary thrombophilia testing NO hereditary thrombophilia testing</div> <div>YES acquired thrombophilia testing YES acquired thrombophilia testing</div>	<div>Minor transient RF</div> <div>≤60 y.o. &gt;60 y.o.</div> <div>YES hereditary thrombophilia testing NO hereditary thrombophilia testing</div> <div>YES acquired thrombophilia testing YES acquired thrombophilia testing</div>	<div>≤60 y.o. male. Positive family history for VTE AND positive family history for thrombophilia</div> <div>YES targeted high-risk thrombophilia testing NO low-risk thrombophilia testing</div>	<div>Positive family history for VTE AND positive familial thrombophilia</div> <div>Pregnancy, oestrogen CC</div> <div>YES targeted high-risk thrombophilia testing YES targeted low-risk thrombophilia testing</div> <div>Hormone replacement therapy</div> <div>YES targeted high-risk thrombophilia testing NO low-risk thrombophilia testing</div>	<div>Unprovoked VTE - paediatric</div> <div>Purpura fulminans, severe VTE</div> <div>YES high risk thrombophilia testing immediately</div>
<div>Persisting clinical RF (e.g. obesity, other chronic inflammatory conditions, not malignancy)</div> <div>≤60 y.o. &gt;60 y.o.</div> <div>YES acquired thrombophilia testing YES acquired thrombophilia testing</div>	<div>surgical transient RF, non-surgical major RF, malignancy</div> <div>NO hereditary thrombophilia testing NO acquired thrombophilia testing</div>	<div>Positive family history for VTE, familial thrombophilia unknown</div> <div>NO thrombophilia testing</div>	<div>Positive family history for VTE, familial thrombophilia impossible to determine</div> <div>Pregnancy, oestrogen CC</div> <div>NO thrombophilia testing</div>	<div>Provoked VTE - paediatric</div> <div>No family history for VTE or thrombophilia</div> <div>NO thrombophilia testing</div>
<div>Thrombophlebitis in varicose vein (superficial, &lt;10cm)</div> <div>NO hereditary thrombophilia testing NO acquired thrombophilia testing</div>	<div>Provoked VTE – women, hormonal</div> <div>Pregnancy, oestrogen CC</div> <div>YES hereditary thrombophilia testing YES acquired thrombophilia testing</div>	<div>No family history of VTE, Positive family history for low-risk thrombophilia</div> <div>NO thrombophilia testing</div>	<div>No family history of VTE, No family history for thrombophilia</div> <div>NO thrombophilia testing</div>	<div>No VTE in patient - Paediatric</div> <div>Positive family history for VTE AND positive family history for thrombophilia</div> <div>YES targeted high risk thrombophilia testing after first year of life</div> <div>NO low risk thrombophilia testing. Counselling in adolescence according to adult thrombophilia practices</div>
<div>Unusual site VTE</div> <div>≤60 y.o. &gt;60 y.o.</div> <div>YES hereditary thrombophilia testing NO hereditary thrombophilia testing</div> <div>YES acquired thrombophilia testing YES acquired thrombophilia testing</div> <div>Consider further diagnostics Consider further diagnostics</div>			<div>History of pregnancy complications</div> <div>NO hereditary thrombophilia testing YES acquired thrombophilia testing</div>	

Definitions:

Hereditary thrombophilia: Low risk hereditary thrombophilia: mutations factor V Leiden, prothrombin G20210A gene mutation; High risk hereditary thrombophilia: deficiencies of antithrombin, protein C, or protein S, homozygous / compound heterozygous FVL/PGM; Acquired thrombophilia: antiphospholipid syndrome (APS, 1 or more of Lupus anticoagulant, anticardiolipin antibodies, and anti-β2-glycoprotein 1 antibodies plus history of thrombotic event and/or pregnancy complications).

Abbreviations:

oestrogen CC (containing contraceptive), RF (risk factor), VTE (venous thromboembolism)