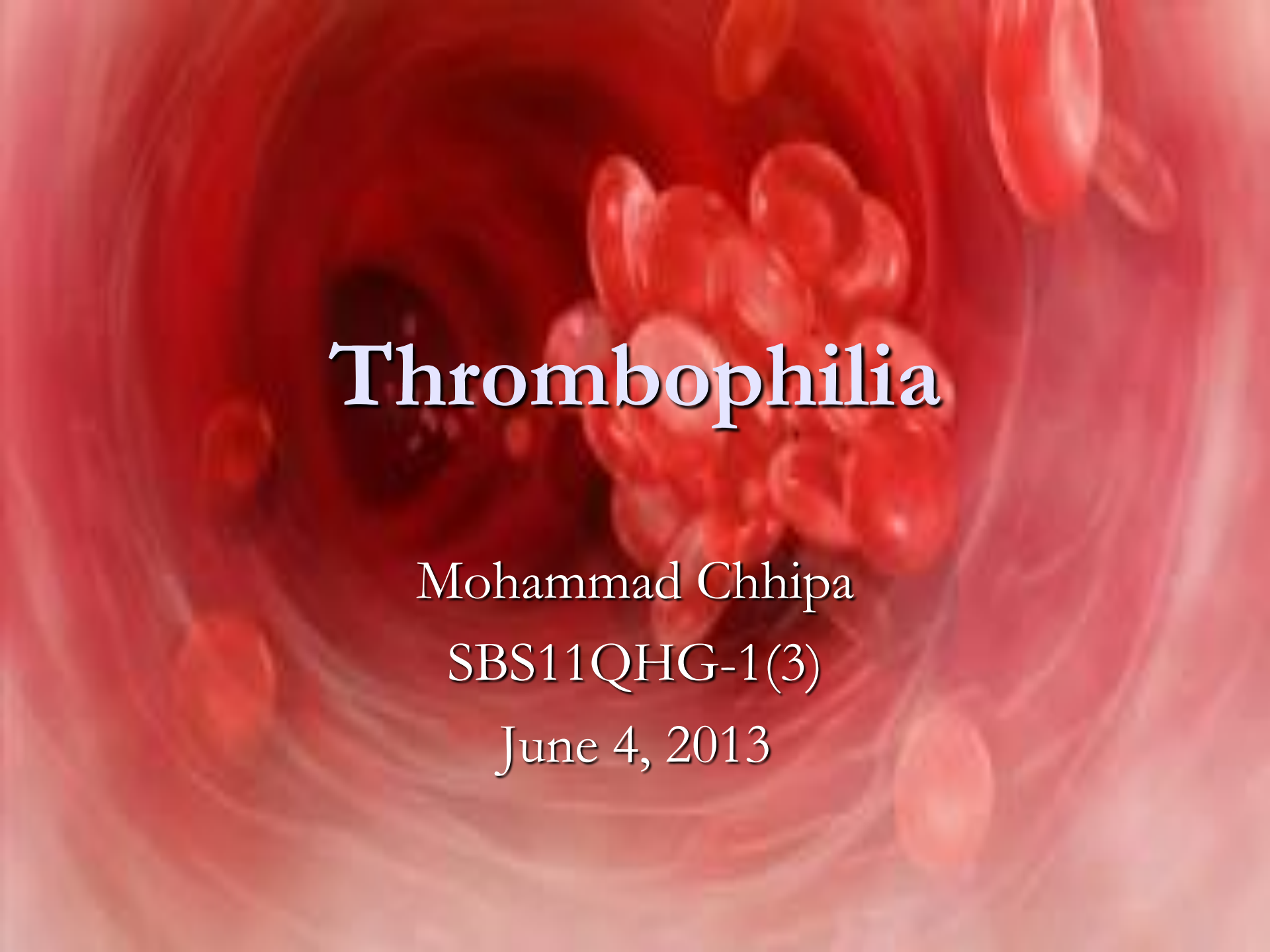


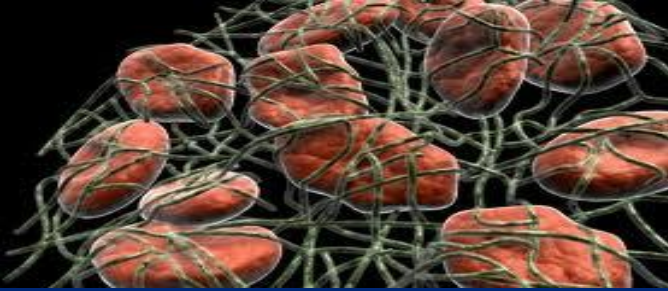
A microscopic view of a blood vessel, showing a central dark thrombus and numerous red blood cells flowing around it. The vessel walls are visible as concentric rings of pinkish-red tissue.

Thrombophilia

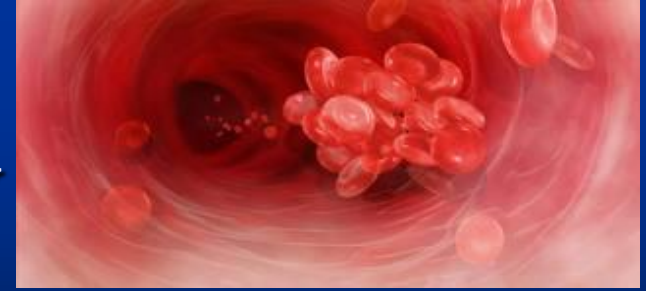
Mohammad Chhipa

SBS11QHG-1(3)

June 4, 2013

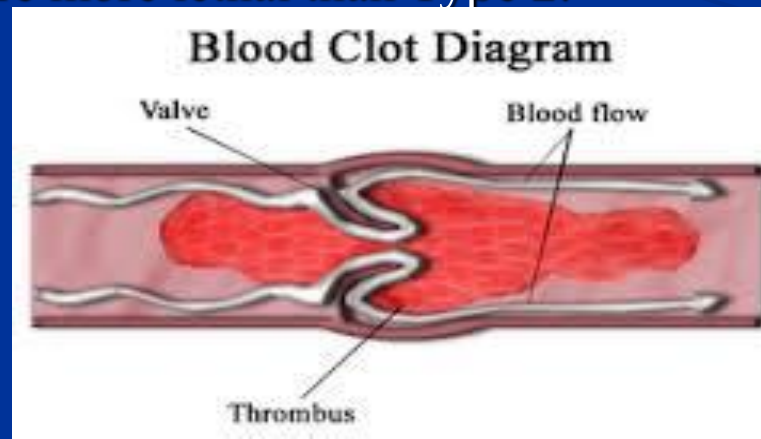
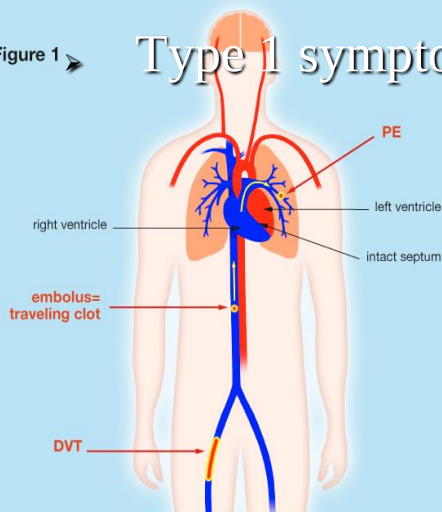


Physiology



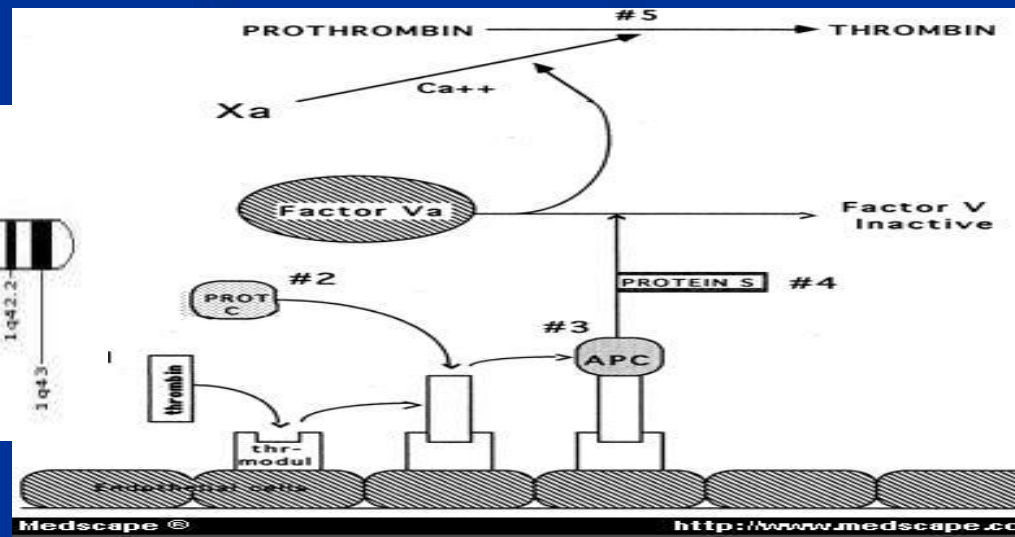
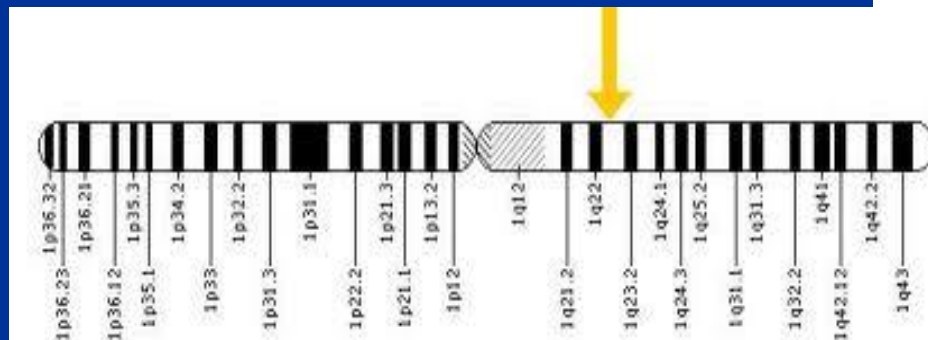
- Affects approximately 0.2% of the general population.
- Most common symptoms include Venous Thromboembolism.
- Targets Circulatory System: specifically the production of the protein Thrombin
- Most common in Northern Europe (Siberia)
- Generally late onsets (most common to 60+ individuals)
- Other symptoms include chest pain, shortness of breathe, blood clots within blood vessels, swelling of veins, and abnormal blood coagulation.

Figure 1 ➤ Type 1 symptoms are more lethal than Type 2.



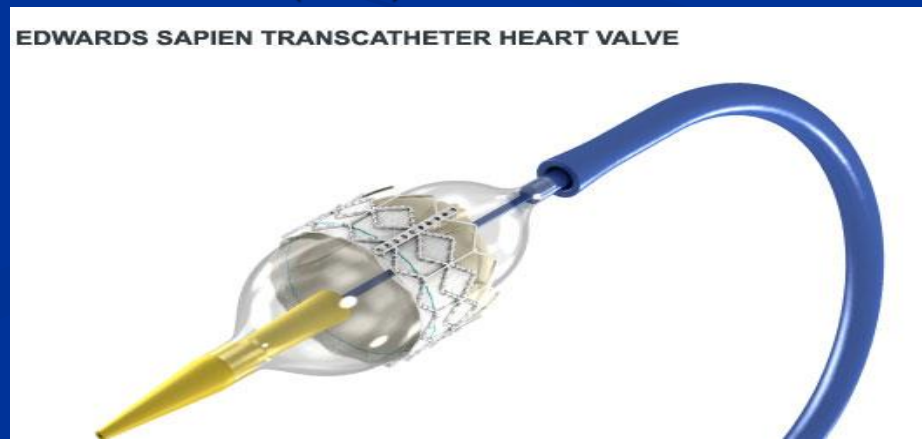
Molecular Cause

- Translocation of Factor V Leiden, located on Chromosome 1, specifically on the F5 gene at position 1691
- Leads to excess Thrombin production; these Thrombin protein will lack Xa receptors.
- Inability to control Thrombin production due to Activated Protein C (aPC) deficiency.
- Autosomal Recessive manner of inheritance pattern.
- Inability for Thrombin to return to its inactivated form, Prothrombin
- Too many blood clots formed as Thrombin activates Fibrin into Fibrinogen.



Treatments/Limits

- No known cure, only treatments for symptoms
- Early detections by blood film examination.
- Edwards Sapien Heart Transcatheter: Prevents blood clogging by expanding heart valves, ex. Sino-atrial valve.
- Leads to high risk of stroke and other heart problems due to insertion of cow heart tissue into human heart.
- Warfarin Tablets.
- Restores anti-coagulation factors by using insect proteins to get rid of emboli in blood stream and get rid of Thrombin.
- Leads to severe bleeding up to high risk of death (11%).



Proposed Cure/Limits

- Because of the excess Thrombin proteins causing emboli to clog blood vessels I will inactivate them into Prothrombin.
- Restore Anti-coagulation factors such as Protein S to that they will counteract Thrombin production. .
- Take mature mRNA from humans that has been spliced and insert into bacteria.
- Use enzyme reverso-transcriptase to put mRNA into bacteria's DNA.
- Once Proteins S has been transcribed the production of Protein S will be cultured and injected into Thrombopliacs.
- Because bloodstream is direct target for protein S, we are safe in terms of the Protein reaching the defected Thrombin molecules.
- Limitations are that the body might reject the foreign Protein S.
- Too much Thrombin might be inactivated, which can cause severe bleeding.

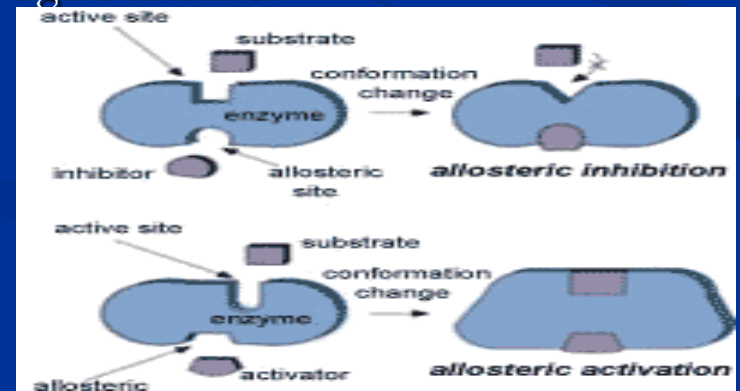


Image on cover and Physiology Page:

https://www.google.com/search?q=thrombophilia&source=lnms&tbm=isch&sa=X&ei=x0WtUbaHtbf4APBm4DABw&ved=0CAoQ_AUoAQ&biw=1366&bih=643#facr=&imgre=va0MRYL9tn-EaM%3A%3BSRtQ8PlYtnHcpM%3Bhttp%253A%252F%252Fwww.kofinasperinatal.org%252Fimages%252FSourceImage%252FThrombophilia%252520Presc-3FD384A2.jpg%3Bhttp%253A%252F%252Fwww.kofinasperinatal.org%252F%3B520%3B260

Images on Physiology Page:

https://www.google.com/search?q=thrombophilia&source=lnms&tbm=isch&sa=X&ei=UbaHtbf4APBm4DABw&ved=0CAoQ_AUoAQ&biw=1366&bih=643#facr=&imgre=2zsr8TOCIIImMM%3A%3BjdlR3fnowu7YM%3Bhttp%253A%252F%252Fclotconnectblogtwo.apps.newmediacampaigns.com%252Fwp-content%252Fuploads%252F2011%2F202%252Ffigure-1.jpg%3Bhttp%253A%252F%252Fpatientblog clotconnect.org%252Fcategory%252Fdiagnosis%252F%3B720%3B576

https://www.google.com/search?q=thrombophilia&source=lnms&tbm=isch&sa=X&ei=rCsUUbOqNIOs0QHBYCoAQ&ved=0CG0QexY&fp=1&biw=1366&bih=643&bav=on.2.or.r_qf.&cad=b&sei=xUmtUz4Mc2s4AO_14GyIbq&facr=&imgre=q0hREMrdq92YGM%3A%3Bu2GxAienqCZc4M%3Bhttp%253A%252F%252Fwww.stoptheclot.org%252Fimages%252Fnatl_other_art%252Facute%252520left%252520leg%252520DVT%253B%252520postthrombotic%252520syndrome%252520right%252520leg.jpg%3Bhttp%253A%252F%252Fwww.stoptheclot.org%252Flearn_more%252Flearn_thrombosis.htm%3B2415%3B3094

https://www.google.com/search?q=swollen+legs&source=lnms&tbm=isch&sa=X&ei=kitUY-rdJcq4AOxzIHYCA&ved=0CAoQ_AUoAQ&biw=1366&bih=643#tbm=isch&q=swollen+legs+thrombophilia&spell=1&sa=X&ei=CEmtUZcuONW14APE8oDgAw&ved=0CE8QvwwUoAA&fp=1&biw=1366&bih=643&bav=on.2.or.r_qf.&cad=b&sei=xUmtUz4Mc2s4AO_14GyIbq&facr=&imgre=q0hREMrdq92YGM%3A%3Bu2GxAienqCZc4M%3Bhttp%253A%252F%252Fwww.stoptheclot.org%252Fimages%252Fnatl_other_art%252Facute%252520left%252520leg%252520DVT%253B%252520postthrombotic%252520syndrome%252520right%252520leg.jpg%3Bhttp%253A%252F%252Fwww.stoptheclot.org%252Flearn_more%252Flearn_thrombosis.htm%3B2415%3B3094

https://www.google.com/search?q=thrombophilia&source=lnms&tbm=isch&sa=X&ei=mhasUciZC6fA4APQYDgCQ&ved=0CAoQ_AUoAQ&biw=1366&bih=643#tbm=isch&sa=1&q=blood+clogs&og=blood+clogs&gs_l=img.3..0l10.31041.32763.0.32888.11.10.0.0.0.180.682.8j1.9.0..0.0.0..1c.1.15.img.7e0xwW_15zM&bav=on.2.or.r_qf.&fp=p72948f606f9b4c&biw=1366&bih=643&facr=&imgre=Y9wflYXPg8nj9M%3A%3BZws8j3ofRyhnGM%3Bhttp%253A%252F%252Fupload.wikimedia.org%252Fwikimedia%252Fcommons%252Fthumb%252F%252F5%252FBlood_clot_diagram.png%252F300px-Blood_clot_diagram.png%3Bhttp%253A%252F%252Fen.wikipedia.org%252Fwiki%252FThrombus%3B300%3B189

[1].Haematolov, Br.. "Thrombophilia in pregnancy: a systematic review.." <http://www.ncbi.nlm.nih.gov/pubmed/16398652>. Pubmed.gov, 2 1 2006. Web. 21 Mar 2013.

2."Thrombophilia Health.N.D." <http://www.patient.co.uk/health/thrombophilia>. BMA Patients Information. Web. 21 Mar 2013.

3.Fuster, Valentin . "Cardiovascular Research." <http://icahn.mssm.edu/research/institutes/cardiovascular-institute>. Mount Sinai, n.d. Web. 20 Mar 2013.

4."Thrombophilia Symptoms." <http://vascularisease.org/thrombophilia/thrombophilia-symptoms/>. Vascular Disease Foundation, 29 3 2012. Web. 21 Mar 2013.

5. Andrea , Luigi. "Thrombophilia Symptoms." <http://www.intechopen.com/books/thrombophilia>. INTECH, 9 11 2011. Web. 21 Mar 2013.

Images on Molecular Cause Page:

https://www.google.com/search?q=factor+vv+leiden&source=lnms&tbm=isch&sa=X&ei=rCCsUZYFYu50AGsloCICw&ved=0CAoQ_AUoAQ&biw=1366&bih=643#tbm=isch&q=factor+vv+leiden+gene&revid=1065700870&sa=X&ei=rSCsUUbOqNIOs0QHBYCoAQ&ved=0CG0QexY&fp=1&biw=1366&bih=643&bav=on.2.or.r_qf.&cad=b&sei=xUmtUz4Mc2s4AO_14GyIbq&facr=&imgre=q7y2kPljYmsRiM%3A%3BXv6vxxDaaamMOM%3Bhttp%253A%252F%252Fghr.nlm.nih.gov%252FdynamicImages%252Fchromomap%252F5.jpg%3Bhttp%253A%252F%252Fghr.nlm.nih.gov%252Fgene%252F5%3B649%3B179

https://www.google.com/search?q=thrombin+pathway&source=lnms&tbm=isch&sa=X&ei=f0mtUceOG4na4AO9IHdYDQ&ved=0CAoQ_AUoAQ&biw=1366&bih=643#facr=&imgre=oyJ8JKhEIqtU6M%3A%3BE3RcTmrkdN9nAM%3Bhttp%253A%252F%252Fimg.medscape.com%252Ffullsize%252Fmigrated%252F415%252F086%252Fsmj2410.06.fig1.jpg%3Bhttp%253A%252F%252Fwww.medscape.com%252Fviewarticle%252F415086_3%3B400%3B461

6. Gayathri, Parasivam"Blood Clotting Conditions""http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/Blood-Clotting-Conditions-Thrombophilias-Bleeding-Conditions1-FS39/at_download/file". Centre for Genetics Education, Web. 15 May 2013.

7. "Protein S Deficiencies and Blood Clots" <http://www.ucdenver.edu/academics/colleges/medicalschoo/centers/HemophiliaThrombosis/resources/Documents/PROTEIN_S_DEFICIENCY.pdf>. Web. 19 May 2013.

Images on Treatments/Limits Page

<http://www.edwards.com/Products/TranscatheterValve/Pages/THVcategory.aspx>

https://www.google.com/search?q=warfarin&source=lnms&tbm=isch&sa=X&ei=c0qrUaalAvOy4AP9sYBw&ved=0CAoQ_AUoAQ&biw=1366&bih=643#tbm=isch&sa=1&q=warfarin+tablets&og=warfarin+tablets&gs_l=img.3..0l4j0i2414.6993.8182.1.8299.11.10.0.1.1.0.131.865.7i3.10.0..0.0.0..1c.1.15.img.zmnZ_WFLY&bav=on.2.or.r_qf.&biwm=bv.47244034.dimg&fp=p72948f606f9b4c&biw=1366&bih=643&facr=&imgre=q1I9Yur5SGnYM%3A%3BzhTXnO8uzbM6fM%3Bhttp%253A%252F%252Fupload.wikimedia.org%252Fwikipedia%252Fen%252F5%252F59%252FWarfarintablets5-3-1.jpg%3Bhttp%253A%252F%252Fen.wikipedia.org%252Fwiki%252FFile%253AWarfarintablets5-3-1.jpg%3B800%3B496

8. Cunha, D., M. Wedro, and M. Conrad-Stoppler. N.p.. Web. 19 May 2013. <http://www.emedicinehealth.com/blood_clots/article_em.htm>.