

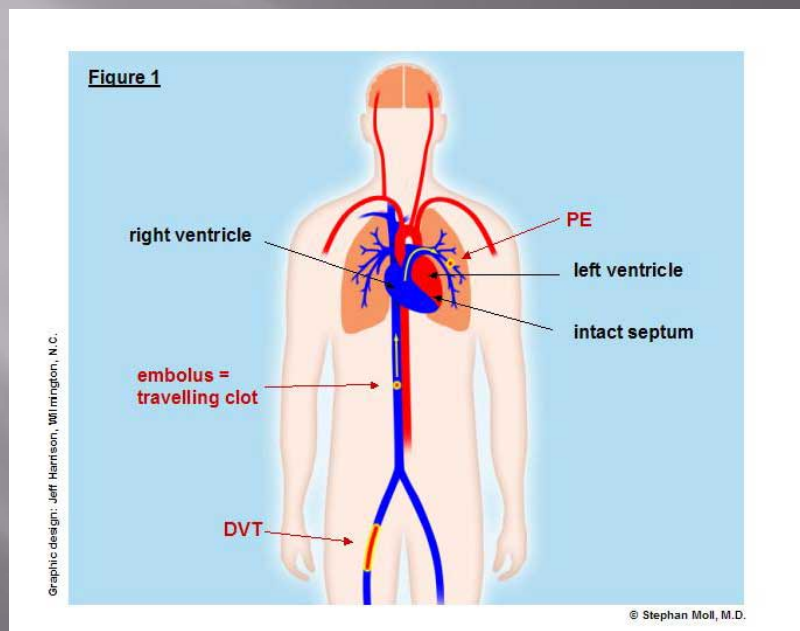
Diagnosticul molecular al mutatiei Leiden a Factorului V la pacientii cu trombofilie

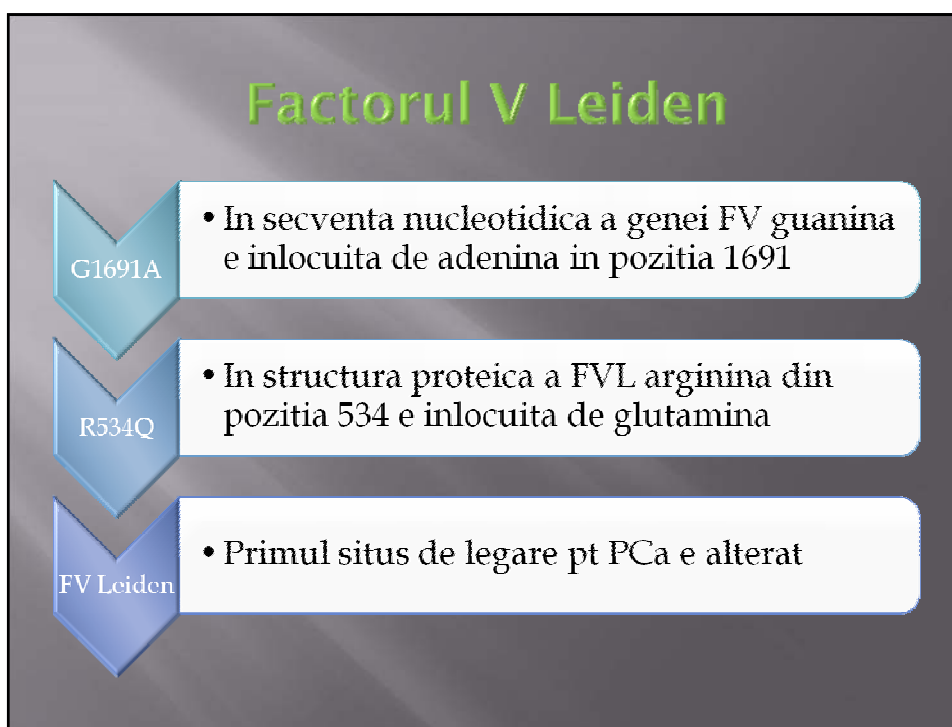
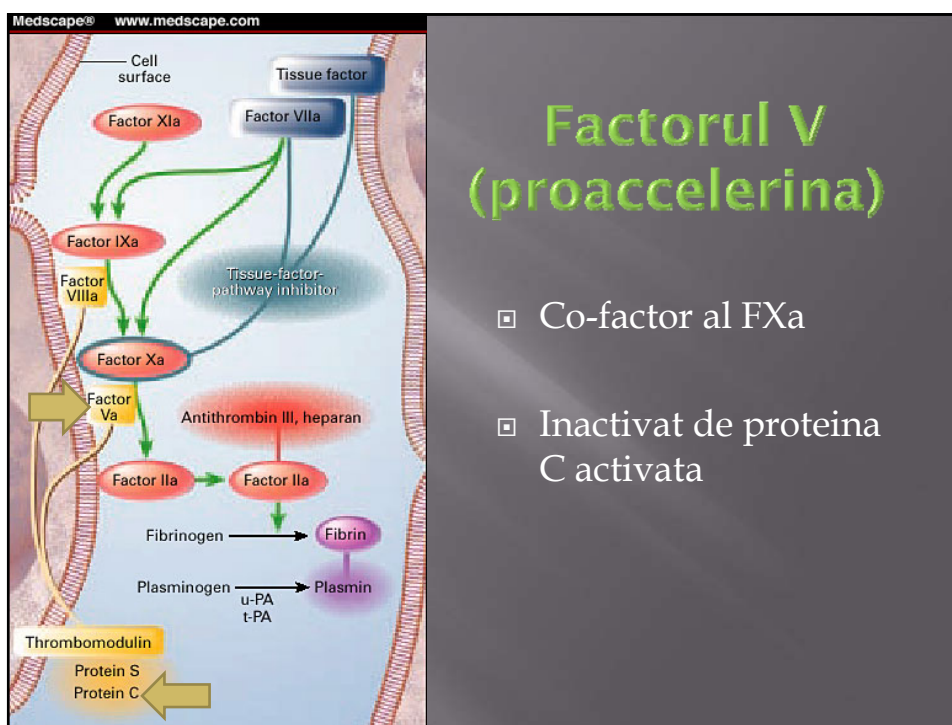
Studenta: Mitroi Gabriela

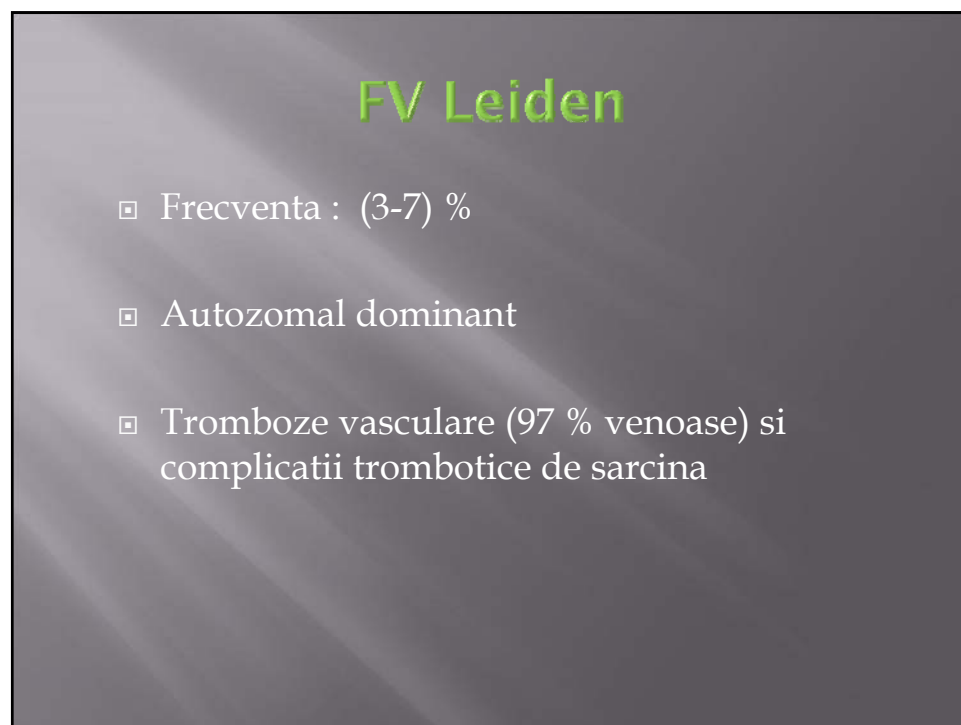
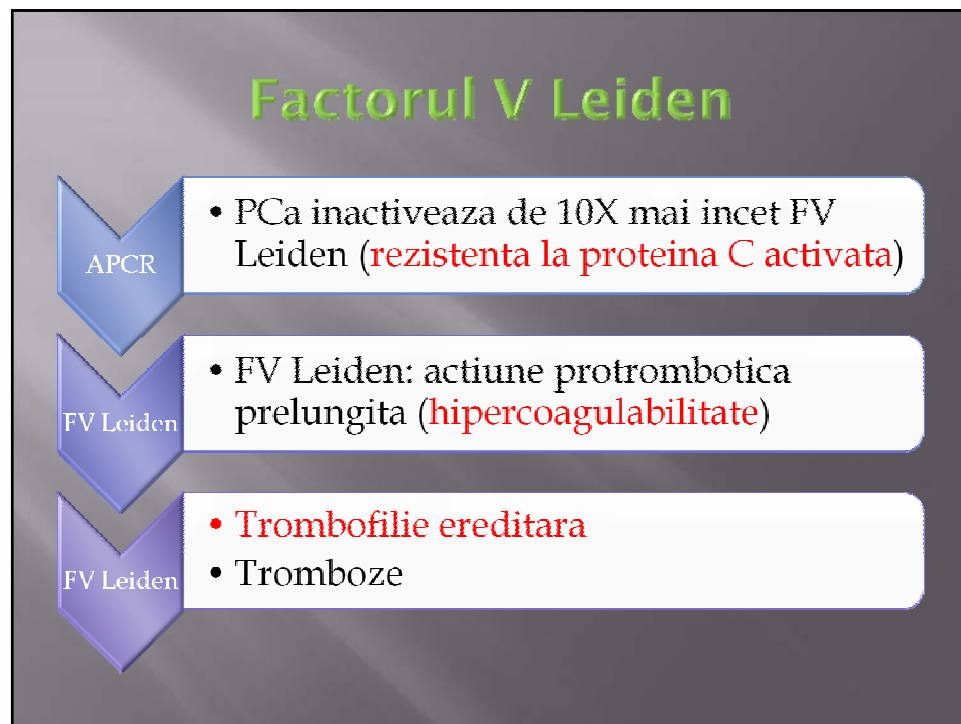
Coordonatori științifici: As. Univ. Rodica Talmaci

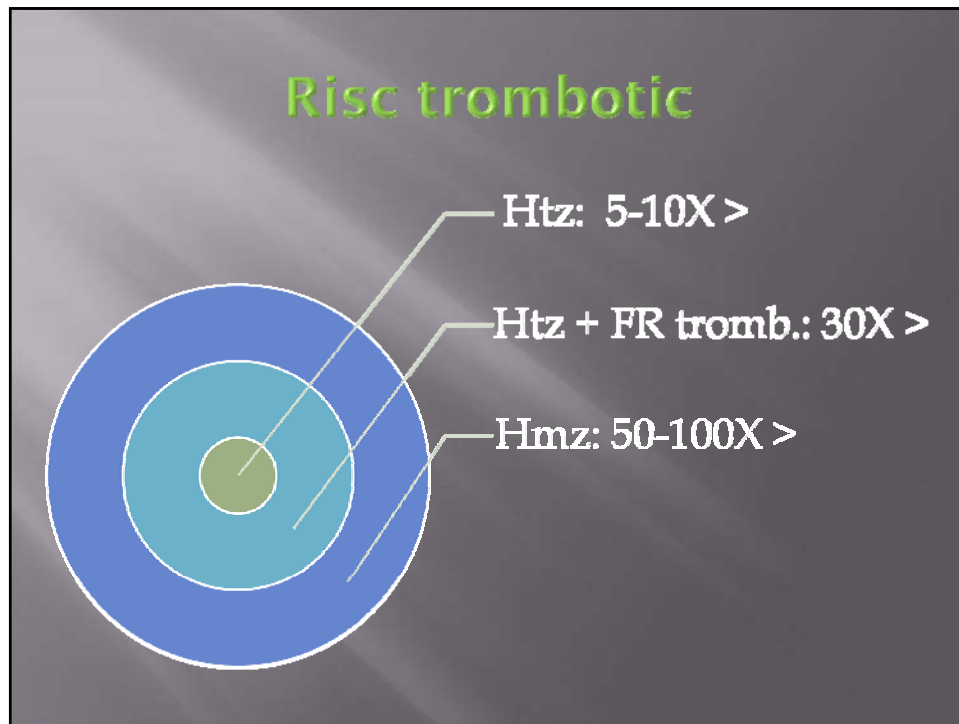
Conf. dr. Daniel Coriu

Institutul Clinic Fundeni, Hematologie









Diagnosticul FV Leiden

- ▣ Investigarea APCR (un aPTT modificat)
ProC Global cut-off 0,8
- ▣ Analiza moleculara: evidentiarea mutatiei la nivelul AND-ului

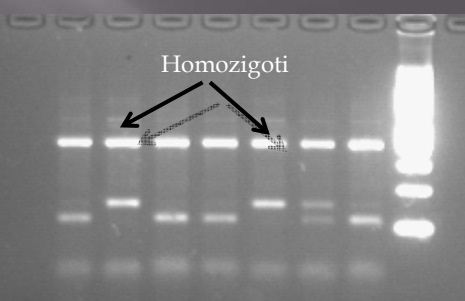
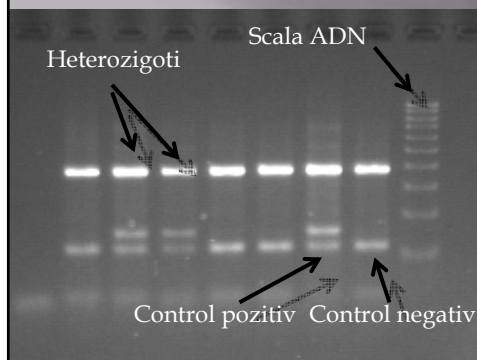
Analiza moleculara

Izolare ADN din sange
periferic

Amplificare prin PCR

Digestie enzimatica

Interpretare rezultate Electroforeza



APCR

- ▣ In (90-95)% din cazurile cu APCR este prezent FV Leiden
- ▣ Alte cauze de APCR:
 1. Sarcina
 2. FV Cambridge
 3. FV Hong-Kong
 4. FV HR2 si HR3

De aceea...

- ▣ APCR: test de screening la pacienti cu trombofilie in vederea analizei moleculare definitive
- ▣ APCR: are valoare negativa predictiva buna – exclude existenta mutatiei

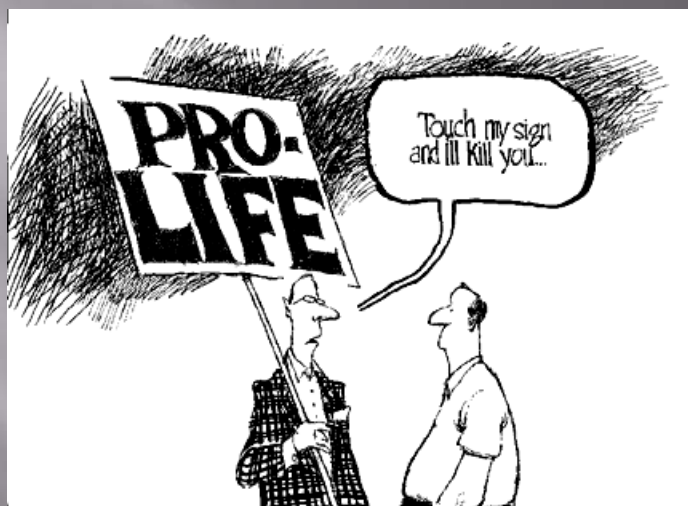
Recomandare

Toti pacientii tineri (<55 ani) cu tromboze ar trebui investigati in vederea evidentierii trombofiliei ereditare

Diagnosticul de trombofilie ereditara este transat de analiza moleculara

5 motive pt analiza moleculara

1. Diagnosticul final al unui eveniment trombotic
2. Diagnosticarea precoce a rudelor de gradul 1
3. Preventie primara: educatie medicala
4. Preventie secundara: terapie anticoagulanta
5. Scaderea morbiditatii si mortalitatii



Va multumesc