Rubinstein-Taybi Syndrome and Keloids

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The problem
Topics

• Scars
• Keloids
• Keloids in RTS
• Study in RTS
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Scars

- end point of tissue repair
- no scars in wounds during pregnancy
- balance between many genes and other factors
Wound healing

- Fibroblasts
- TIMP
- TGF-β
- VEGF
- PDGF
- MAP
- MMP
- Scars

Wound healing
Normal wound healing

3 phases
- inflammation
- proliferation
- maturation/ remodeling
Topics

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Definition

• continuous / invasive scar formation beyond boundaries of the original wound

• edges often red and pruritic

pm differences with hypertrophic scar
Characteristics

• in fact benign tumor
• unique to humans
• only in genetically susceptible individuals
• not at every body site
keloid

Origin

- after surgical incision
- after small trauma (e.g., ear piercing)
- after insect bites
- after burns
- after tattoos
- spontaneously (?)
keloid
keloid
Why?

- familial clustering
- concordance in identical twins
- genetic markers
- immunological changes
Where?

• ear lobes
• shoulders
• chest
• upper back
RTS and keloid
When?

• at any age
• especially between 10 and 30 yr
How often?

- women >> man
- puberty ↑
- pregnancy ↑
  → hormones!
- ethnicity
  - african-americans ↑
  - hispanics ↑
  - asians ↑
Complaints

- itching
- pain
- cosmetic
- limitation joint mobility
Quality of live

- severe impairment
- itching/ tenderness/ pain/sleep disturbance
- physical deformity
- loss of self esteem/ stigmatisation

keloid
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Keloids in syndromes

- very infrequent
  - Goeminne syndrome
  - Rubinstein Taybi Syndrome

n=5 worldwide!
RTS and keloid
How often?

- 20-25%
- spontaneous (?)
- Complaints of itching and pain
Complaints

- itching
- pain
- infections

RTS and keloid
Why in RTS?

- cause RTS mutations in CBP or p300
- keloids only in CBP changes
- keloids because of CBP changes?
Therapy

- many tried
- none really effective
- high recurrence rate
Types of therapy

- compression
- steroid injections
- cryosurgery
- radiation
- 5-Fluorouracil
- Interferon
- laser
- brachytherapy
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Goal

If we understand why and how keloids arise, we may be able to offer better treatment or even prevent keloids.
Stop keloid formation

- wounds / trauma
- Keloid

A, B, C
Who can participate?

• everyone with RTS and keloids
• DNA change confirmation
  pm if needed DNA study done
Where?

- Netherlands
  - Leiden
    - molecular genetics Dr Dorien Peters
  - Amsterdam
    - plastic surgery Dr Annekatrien van de Kar / Prof Chantal van der Horst
    - genetics Prof Raoul Hennekam

- UK
  - London
    - genetics Dr Adam Shaw
study in RTS

How

- Sign form
- RTSandkeloid@gmail.com
First part

- 3 questionnaires
  a. general info / RTS
  b. keloids
  c. itching
- clinical pictures
  - now
  - send by e-mail
Second part

- one visit to hospital
- blood sample if gene change unknown
- 2 keloid biopsies

pm refund travel costs
Third part

- uncertain at present !!
- only if changes confirm ideas about cause

- local medication
  - treat one part of keloid
  - control: other of keloid
- after 6m / 1yr 1 keloid biopsy
So

- no treatment now
- study will take time
- WE WILL NEED YOUR HELP!