Case 4: clinical data

- 26 year old woman
- Multiple painful papules (adolescence)
- Left part of the body
- Arm, neck and leg
Case 4: Biopsy of a papule

- Intra-dermal proliferation (whole dermis)
- Bundles of spindle-shaped eosinophilic cells with elongated nuclei with blunt ends
- Interlacing and whorled pattern
- Flattened epidermis
- Stroma: scarce, slightly fibrous
Case 4: Biopsy of a papule

Well circumscribed lesion

Smooth muscle cells: no atypia nor mitosis

Histological diagnosis: CUTANEOUS LEIOMYOMA
Case 4: clinical data

- Father: same papules lumbar left

- OUR PATIENT:
  - Uterine leiomyomatosis (haemorrhage)
Case 4: final diagnosis

Association of multiple cutaneous leiomyoma, uterine leiomyoma with multiple leiomyoma in her father

**Familial cutaneous leiomyomatosis**
*(Reed syndrome)*

**Segmental type**

Characterized by uterine and cutaneous leiomyoma (1954)

More recently association with papillary renal-cell carcinoma
Case 4: Reed syndrome

- Rare inherited affection (165 families; 15 France)
- Autosomal dominant
- >10 cutaneous leiomyoma
- 1: histologic confirmation
Case 4: Reed syndrome cutaneous lesions

• Leiomyoma:
  – Smooth muscle cell benign tumor derived from the arrector pili muscle (piloleiomyoma)
  – Can exist outside Reed syndrome (solitary)
  – Reddish-brown papulonodules++, plaques
  – Asymptomatic or painful (pressure, cold)
  – Variably distributed or dermatomal
  – Female preponderance
  – Face, back, extensor surface of extremities
Case 4: Reed syndrome
cutaneous leiomyoma

- Typical histological aspect
Case 4: Reed syndrome extracutaneous lesions

- Women: 100% **uterine Leiomyomatosis** (diagnosis at 30 y old) pain, hemorrhage, fertility trouble, spontaneous miscarriage
  
  Uterine LMM leads to hysterectomy < 40 Y old

- **Renal carcinoma** (adenocarcinoma papillary type II)

- Rare uterine Leiomyosarcoma (premenopausal) breast or bladder carcinoma, renal cysts (our patient)
Case 4: Reed syndrome

Genetic data

- Autosomal dominant
- Hereditary leiomyomatosis related to renal cell cancer syndrome (HLRCC syndrome)
- Result from heterozygous germline mutations in the Fumarate Hydratase gene (Kreb’s cycle)
- Gene localized on chromosome 1q42-44
- Act as tumor suppressor gene
- No definite association between the site or type of mutation and risk of papillary renal-cell carcinoma
Case 4: Reed syndrome Familial leiomyomatosis

- To recognize cutaneous lesions: multiple leiomyoma
- To detect
  - uterine leiomyomatosis
  - Occult renal malignancy

*Complete history, physical examination, gynecological examination and echography abdominal tomodensitometry*

- Familial examination, genetic counseling