Autoantibodies: The mystery revealed

Connective Tissue Diseases

Houssem ABIDA & **Yasmina OUERDANI**
Faculty of medicine of Tunis *Faculty of medicine of Sousse*

- First description: **1987** in a patient with Systemic sclerosis (SSc)

Target antigen

- A 90 kDa protein identical with **the human upstream binding factor (hUBF)**
- Binds to the promoter of the ribosomal RNA gene and **increases the transcriptional activity of RNA polymerase I**

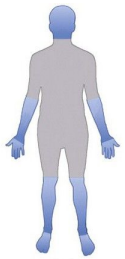
Family: SSc associated antibodies

Anti-NOR90 antibodies

Main disease: Systemic sclerosis

The prevalence in patients with SSc: 3,3 % (44 among 1318 patients in the EUSTAR cohort)

Clinical features in Systemic Sclerosis:

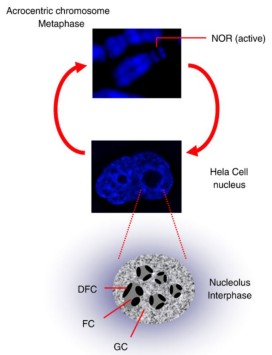


- Female predominance
- **Skin involvement:**
  mostly limited cutaneous systemic sclerosis
  lower modified Rodnan Skin Score
- Less frequent gastrointestinal symptoms
- Less vasculopathy symptoms

⇒ Might be a marker of milder forms of SSc

Also suggested:

- Association with **Interstitial lung disease (ILD)**
- Higher prevalence of cancer
- Association with **Sjogren syndrome** (Japanese cohort)



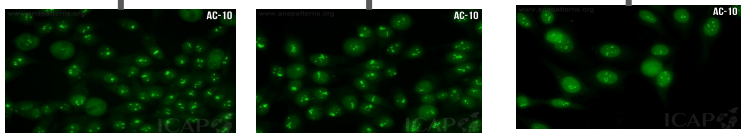
Screening technique: Immunofluorescence on HEp-2 cells

- In interphase cells**, fluorescence is** densely distributed but distinct grains seen in the nucleoli
- In metaphase cells, up to 5 bright pairs of the nucleolar organizer regions (NOR) can be seen within the chromatin body
- The cytoplasm of mitotic cells may be slightly positive

It should be distinguished from other SSc target antigens, particularly the ScL-70 and RNA polymerase I patterns.

→ **Lack of specificity**

IIF pattern on HEp-2 cells: Punctate nucleolar



Confirmation techniques:

- **Line/dot blot immunoassays** Sensitivity: 3,9% Specificity: 99%
- **ELISA** Sensitivity: 91% Specificity: 99%

References