Clinical Case Presentation

ESIM Winterschool Saas Fee 2013

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Migrant Care Program

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The Patient

- Male, 24 years old
- Armenian, Asylum Seeker
- Comes alone to Switzerland in August 2011 to undergo surgery?

The Patient

- Abnormal gait at 6 y.o, wheelchair at 10 y.o, notion of arthrodesis of left hip for an unknown reason
- Associated symptoms: Intermittent fever, chronic diarrhea, rectorragia, ulcerations of mouth and genitals, polyarthritis, loss of weight and appetite, night's sweatings
- Physical examination: weight 38 kg (stable), height 135 cm (BMI 20.9 kg/m2), BP 110/70 mmHg, HR 70/mn, T 37°
- Abdomen: soft, tender diffusely, no sign of of peritonism, no organomegaly, normal sounds
- Right Hip fixed in external rotation, left hip fixed at 0°
- Rest: unremarkable

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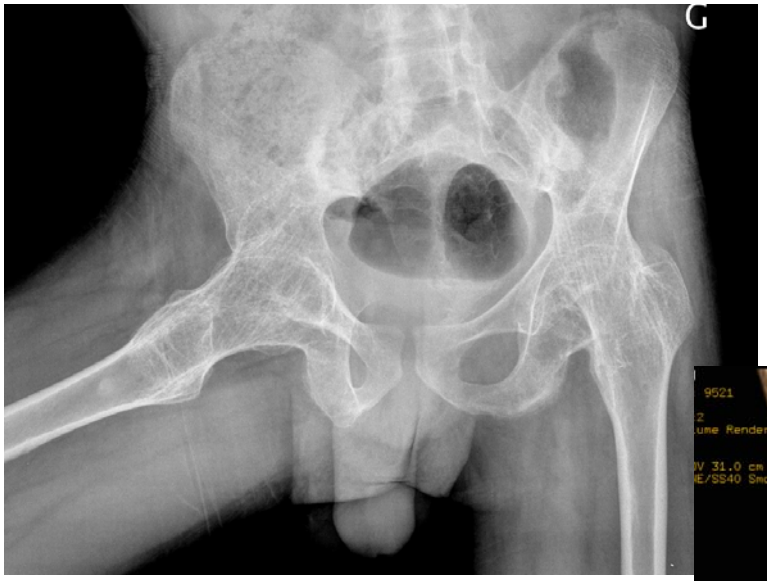
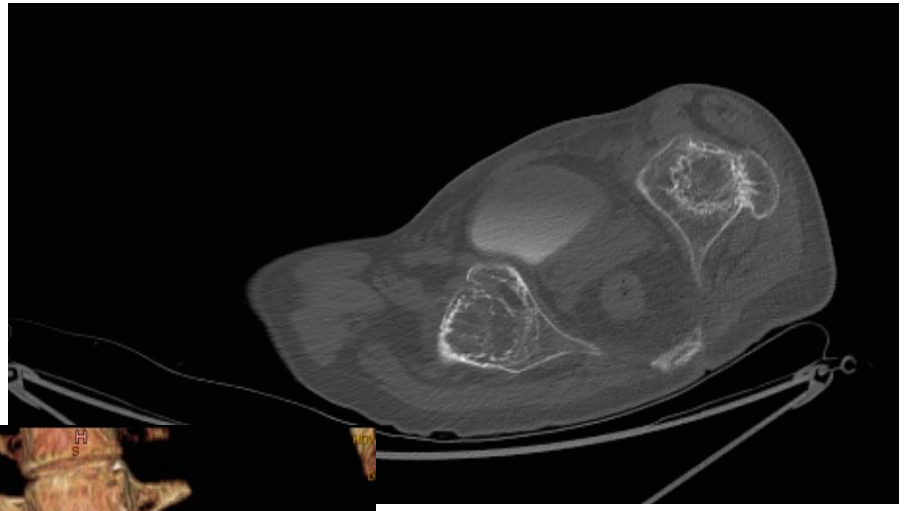
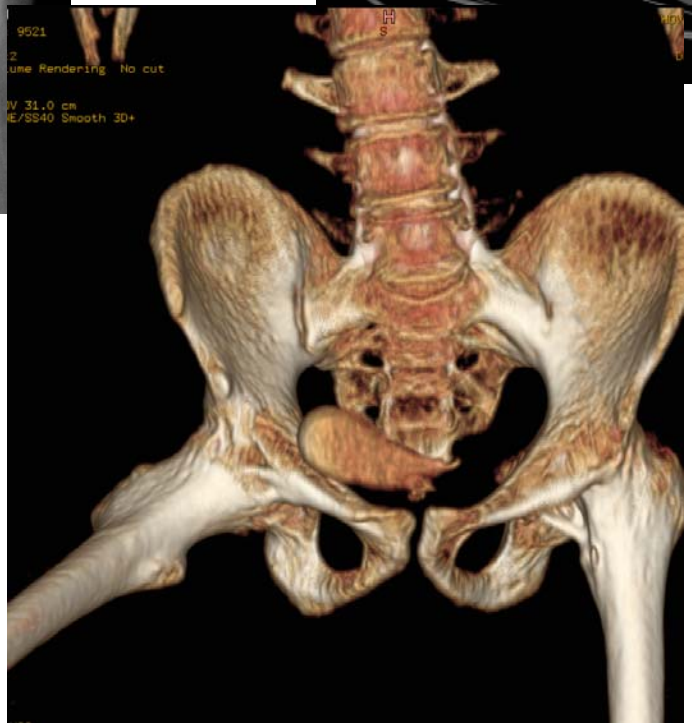
Radiologic exams







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Differential diagnosis?

Pott's disease (Bone Tuberculosis),

Crohn's disease (regional enteritis),

Ulcerative colitis (UD)

Behcet,

NSAID-induced colitis,

Whipple's disease,

lymphoma…

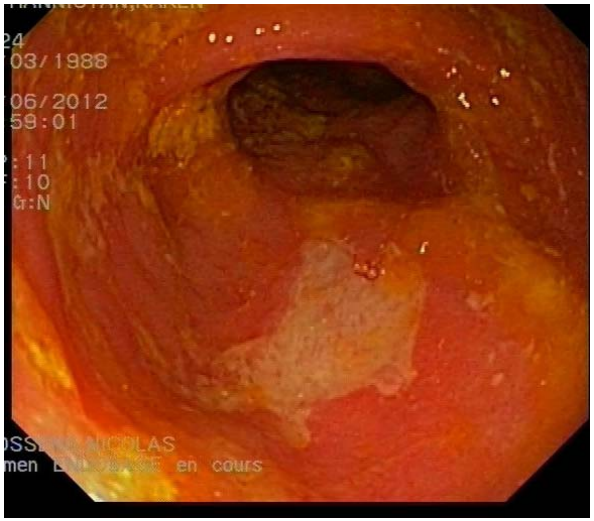
Further investigations

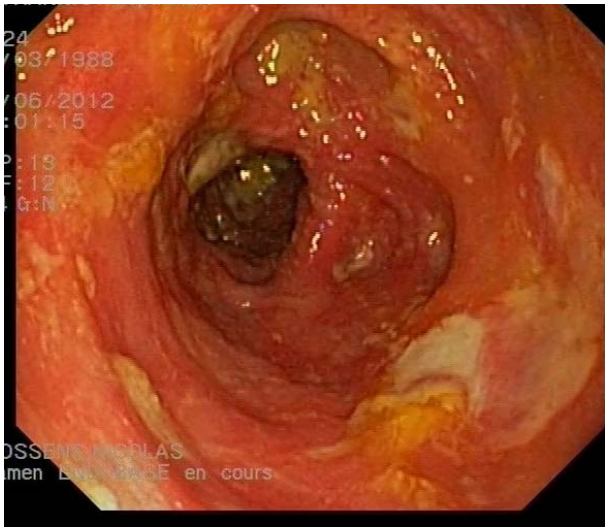
Laboratory:

- Blood count: Microcytic Hypochromic Anemia with hyporegeneration (Hb 110g/l) , no leukocytosis
- Inflammatory markers: ESR 80 (mm/h 0-10), CRP 140 (mg/l 0-10),
- Vitamin D: 9 (nmol/l, > 75)
- Stools and urine culture: negative, no parasites in stools
- HLA B27 and B51: negative
- Interferon γ-Test (T-Spot®.TB): negative
- Immunology: rheumatoid factor, anti-cyclic citrullinated peptide antibodies (anti-CCP), acetylcholine receptor antibody, Anti-neutrophil cytoplasmic antibodies (ANCAs), Antinuclear Antibody Test (ANA): negative
- Immunoglobulins: normal
- HIV and hepatitis: negative
- Flow cytometry: normal

Endoscopy

- Colonoscopy: inflammation of mucosa (skip lesions), polyps of inflammatory aspect
- Gastroscopy: atrophy of duodenal mucosa
- 2nd Colonoscopy: multiples ulcerations in ascendant colon and rectum (progression vs 1st colonoscopy)
- Histology: inflammatory lesions





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Differential diagnosis

Chrohn, NSAID Colitis, Behcet,

Genetic Testing: Two mutations of the MEFV gene (Met680Ile + Met694Val)

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Familial Mediterranean fever (FMF)

- Transmission: autosomal recessive, incomplete penetrance
- Epidemiology: most prevalent in individuals of Sephardic Jewish, Armenian (carrier rate 1:7, disease 1:500), Turkish, Arab descent
- Clinical: first attack in early childhood in 65%, sporadic, paroxysmal attacks of fever and serosal inflammation (pain), 1-3 days, spontaneaous resolution
- Long-term complications: Secondary (AA) amyloidosis
- Diagnostic : typical clinical manifestations, positive response to colchicine therapy, genetic testing, exclusion of other illnesses (one mutation + typical clinical manifestations)

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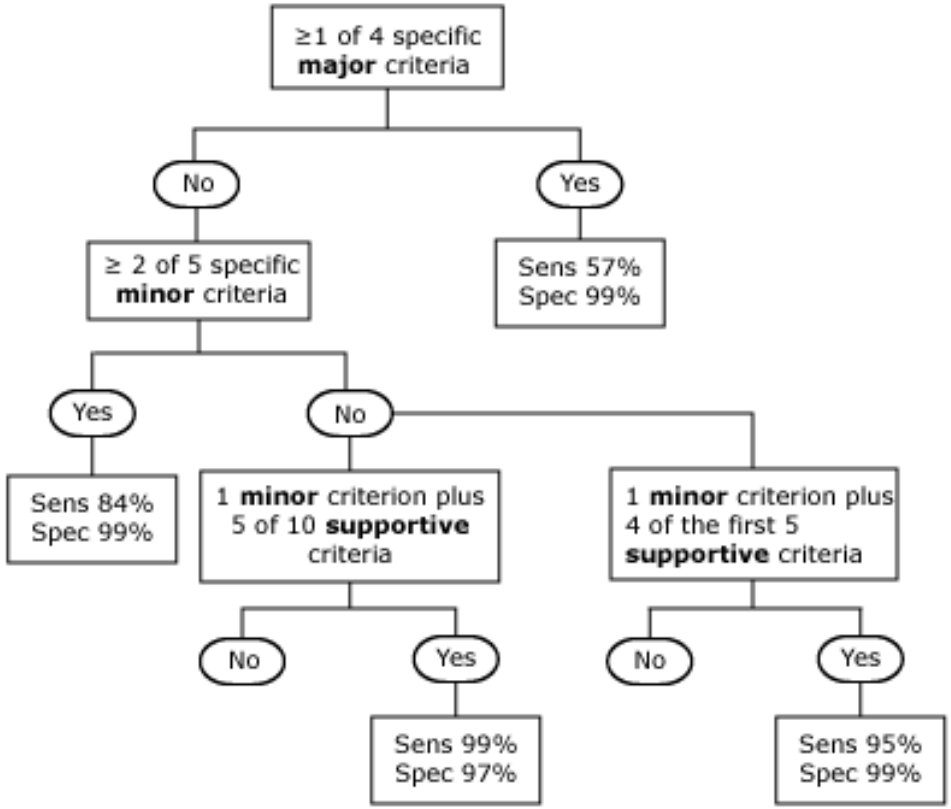
Detailed criteria for the diagnosis of familial Mediterranean fever

Major criteria
Typical attacks
1. Peritonitis (generalized)
2. Pleuritis (unilateral) or pericarditis
3. Monoarthritis (hip, knee, ankle)
4. Fever alone
5. Favorable response to colchicine
Minor criteria
1-4. **Incomplete** attacks involving one or more of the following sites
1. Abdomen
2. Chest
3. Joint
4. Exertional leg pain
Supportive criteria
1. Family history of FMF
2. Appropriate ethnic origin
3. Age <20 years at disease onset
4-7. Features of attacks:
4. Severe, requiring bed rest
5. Spontaneous remission
6. Symptom-free interval
7. Transient inflammatory response, with one or more abnormal test result(s) for the white blood cell count, erythrocyte sedimentation rate, serum amyloid A, and/or fibronogen
8. Episodic proteinuria/hematuria
9. Negative laparotomy or removal of normal appendix
10. Consanguity of parents

Adapted from: Livneh A, Langevitz P, Zemer D, et al. Arthritis Rheum 1997; 40:1879.

Classification tree using the detailed criteria for the diagnosis of familial mediterranean fever



%: percent.

Adapted from: Livneh A, Langevitz P, Zemer D, et al. Arthritis Rheum 1997; 40:1879.

Familial Mediterranean fever (FMF)

- Treatment : prevention of acute attacks and prevention of the development and progression of amyloidosis
  - Colchicine
  - Colchicine non responders: Interferon alpha, infliximab, thalidomide, etanercept, anakinra

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The patient - social

- 10.2012: defined as not eligible
- 11.2012: Appeal due to the medical condition. Federal Office for Migration answer: „Colchicine can be found in Armenia“
- Second Appeal in progress: Colchicine is not working. Infliximab is needed. Difficult to find in Armenia and very expensive. Orthopedic surgery could be considered only after reducing the inflammation

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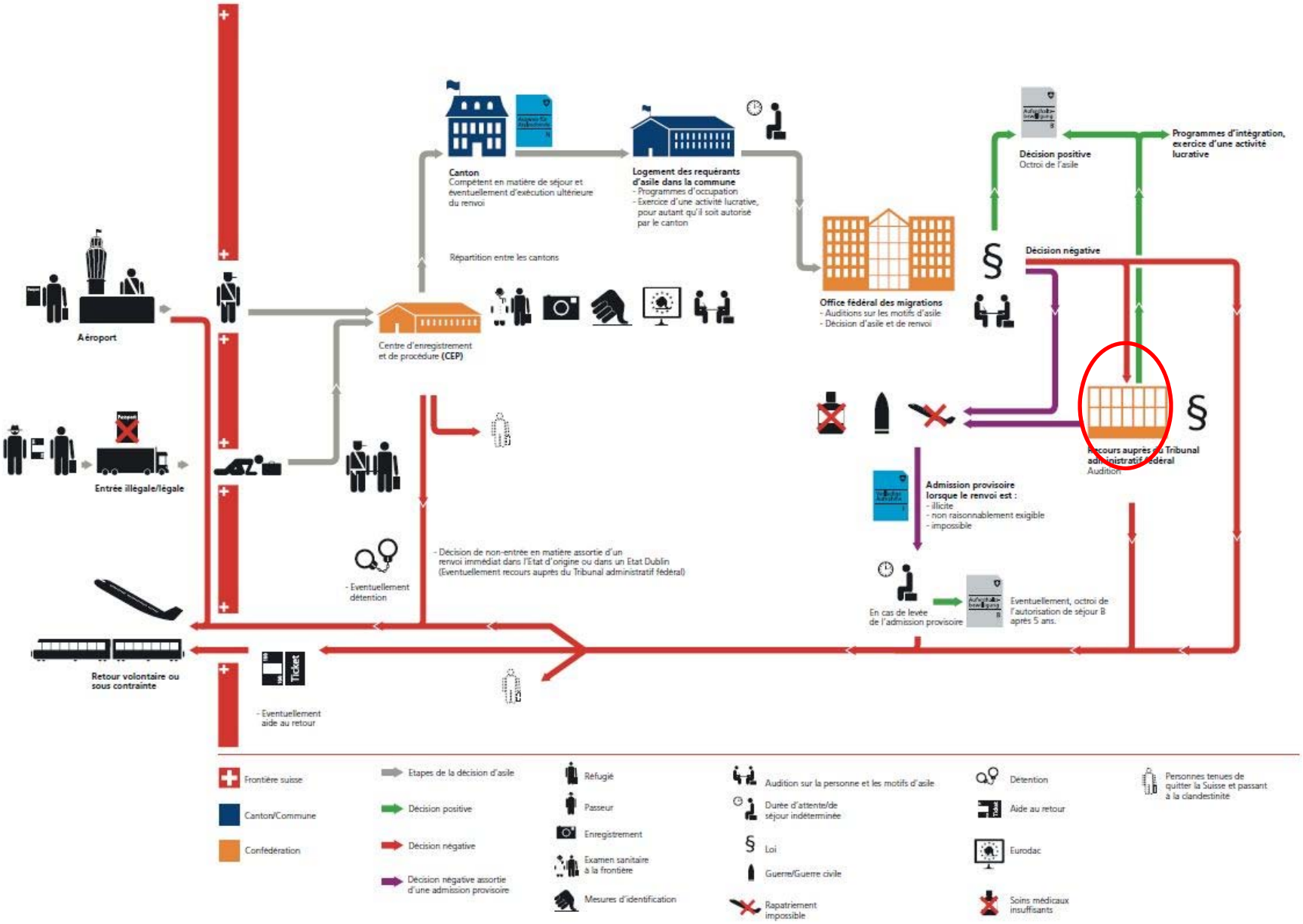

Key points - FMF

- Ethnic predisposition (does not rule out)
- Diagnostic: clinical + genetic + treatment
- Colchicine is a simple and safe treatment
- Several approaches for colchicine non responders

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