Clinical Case

Current History

- Female 41 y
- Smoker, 8-15 cigarettes per day
- No diabetes, BMI 30,8.
- Abscesses of the skin for several years
  - Lesions are usually 1-2 cm in size, usually treated with herbal ointment, no surgery or systemic antibiotics.

Previous History

- No relevant other infectious episodes
- Lower abdominal pain of unknown origin for about 1/2 year
  - Colonoscopy about 8 years ago
  - Laparoscopy 3 years ago
  - GYN exam
- Osteoarthritis of the knee with occasional swelling on evening.
- Migraine

Basic Diagnostics

- CBC, autoimmunity panel and Urinalysis were normal
- Swab of the nasal mucosa was negative
- CRP was not elevated

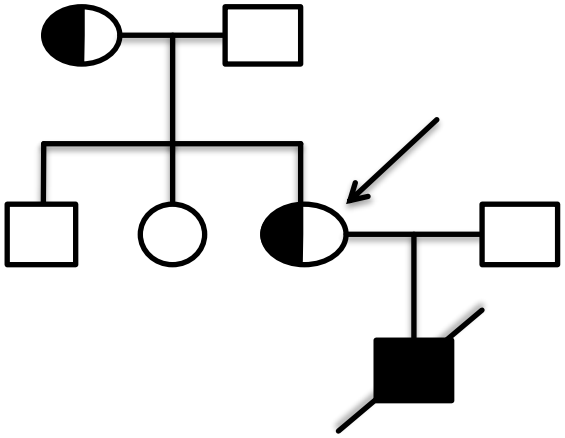
Question 1

- Recurrent skin abscesses in general population:
  - Can be present also in the absence of any predisposing condition
  - Are always linked to an underlying condition
  - Should be considered as a red flag for a PID
  - Are never associated to external social and psychological factors

Question 2

- What other missing information would increase the suspect of a Primary Immunodeficiency?
  - Genetic testing
  - Family history
  - Bacterioscopic culture of the lesion to identify a specific pathogen.

Family History

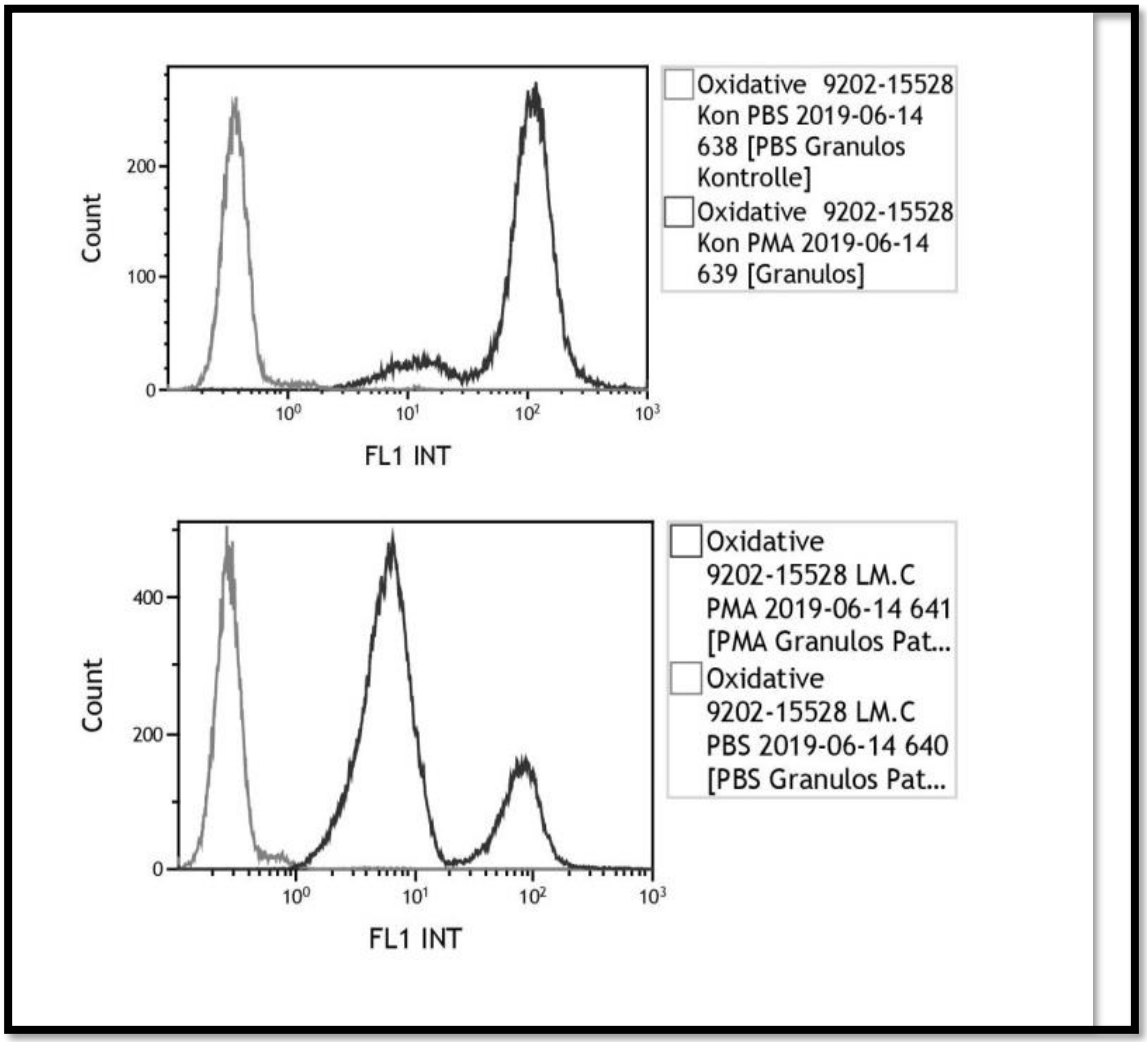


- 09/2008 Carrier for X-CGD [Chronic granulomatous disease] heterozygous Mutation c.1152-2A>T in CYBB (MVZ Humangenetik Uni Dresden).
- An affected son died due to complication of HSCT (2013),
- Patient's mother is also a carrier. One unaffected brother and a sister (carrier status unknown)

Question 3

- what would you expect as a result of the oxidative burst test ?
  - A double peak on neutrophils staining by DHR
  - A single peak on neutrophils staining by DHR
  - A complete absence of any residual activity

Advanced Diagnostics



CONTROL

PATIENT

Question 4

- Which level of Oxidative Burst by neutrophils would be protective against infections?
  - Residual function of 5-10% is generally considered as protective
  - Residual function of 20% is generally considered as protective
  - Residual function of 50% is generally considered as protective
  - There is no consesus on the level of residual function required

Question 5

- What would be the appropriate follow-up for this patient?
  - Re-assess the patient in 6 months to evaluate the clinical picture
  - Herbal ointment to relieve the symptomatology
  - Antibiotic and Antifugal prophylaxys
  - HSCT

Future Course of Treatment

- **How much oxidative function is required for protection against recurrent abscesses?**
  - Carriers with CGD-type infections median about 8%DHR1 (0.06% to 48%)
  - only autoimmune /inflammatory manifestations median 39%DHR1 (7.4%to 74%)[1]
- **Should the patient's clinical picture be considered as an immunodeficiency?**
- **Recommendation for antibiotic prophyaxis?**
- **Recommendation for antifungal prophylaxis?**
- **Definition of the natural history of carriers?**
  - **Infections**
  - **Autoimmunity**
  - **Late onset of clinical manifestations**