Family testing offers a proactive approach to identifying symptomatic X-linked carriers of chronic granulomatous disease (CGD)

PATIENT PROFILE:

Name: Markeyah | **Sex:** Female | **Date of birth:** January 29, 1995

Age at confirmed carrier status: 5 years | **Inheritance pattern:** X-linked

FAMILY HISTORY OF CGD SPANNING 5 GENERATIONS

- Markeyah and her mother are both symptomatic X-linked carriers of CGD confirmed by dihydrorhodamine (DHR) testing
- Her mother experienced multiple boils that required surgical removal, and she was recently diagnosed with lupus
- Multiple male relatives have been diagnosed with CGD, including her brother and 2 deceased cousins

2000: Ringworm infection (tinea capitis)

- Red, itchy, circular rash on the scalp that may have been acquired from a daycare setting
- Pediatrician prescribed an oral anti-infective and topical steroid, but infection was **refractory to treatment for approximately 8 weeks**
- Sought medical attention from her brother's infectious disease (ID) doctor, who **suspected CGD due to family history**
- Due to inadequate response to oral treatment, Markeyah was **hospitalized for 2 weeks** until infection resolved with an **intravenous anti-infective**

2000: DHR testing confirmed X-linked CGD carrier status

- ID doctor ordered a DHR test that confirmed carrier status
- Results revealed a neutrophil oxidative burst pattern consistent with a **symptomatic X-linked carrier at increased risk of infection**

2008: Recurrent skin abscesses

- Developed recurrent axillary and inguinal abscesses caused by the bacterium *Staphylococcus aureus*
- **Antibiotic treatment was insufficient** and required surgical drainage each time
- ID doctor recommended using chlorhexidine wash to help prevent future skin complications

Today:

- Markeyah sees a primary care physician to monitor her boils, a dermatologist for CGD-related eczema, and an ID doctor for **periodic DHR testing**
- She has 2 sons, 1 of whom was born with CGD; both were tested for CGD through DHR testing at 2 weeks old

Family testing enables proactive management of CGD. DHR tests can inform symptomatic X-linked carriers about their risk of infection.

This case study is the experience of 1 individual, and results may vary.

AMGEN Rare Disease

CGD pathways