Non-specific symptoms ultimately identified as autosomal recessive (AR) chronic granulomatous disease (CGD)

PATIENT PROFILE:

Name: Melanie | **Sex:** Female | **Date of birth:** July 12, 1972

Age at diagnosis: 47 years | **Inheritance pattern:** AR

1970s–2000s: A diverse spectrum of symptoms

- History of **chronic fatigue** and **intermittent fevers** starting from childhood
- A persistent pattern of methicillin-resistant *Staphylococcus aureus* (MRSA) infections resulting from minor abrasions and requiring intravenous vancomycin
- Affected ability to work, often resulting in medical or disability-related leave

2010s: Multiple misdiagnoses

- An internist evaluated **escalating symptoms**, ranging from extreme fatigue to body pain, and initially diagnosed Melanie with several non-specific conditions (fibromyalgia, chronic fatigue syndrome) before referring out to an oncologist
- An oncologist completed additional testing with inconclusive results

2019: CGD is confirmed and prompts medical management

- An internist referred Melanie to an allergist/immunologist who suspected CGD
- **Dihydrorhodamine testing** results revealed a neutrophil oxidative burst pattern **consistent with AR-CGD**
- The allergist/immunologist prescribed immunomodulatory therapy and trimethoprim/sulfamethoxazole; an antifungal was not included due to a lack of history for fungal infections
- **No serious* infections or hospitalizations were reported in the first year**

2021: Recovery from an infection

- A knee abrasion led to cellulitis and required surgical removal of the infection

TODAY:

Melanie is on **immunomodulatory therapy and an antibiotic** to manage CGD and has **not had a serious* infection since 2021**. She sees an internist and allergist/immunologist regularly to monitor any changes related to CGD.

This case study is the experience of one individual, and results may vary.
*Serious infection is defined as a clinical event requiring hospitalization and/or intravenous antibiotics.

AMGEN Rare Disease

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CGD pathways