A Late Diagnosis of Chronic Granulomatous Disease

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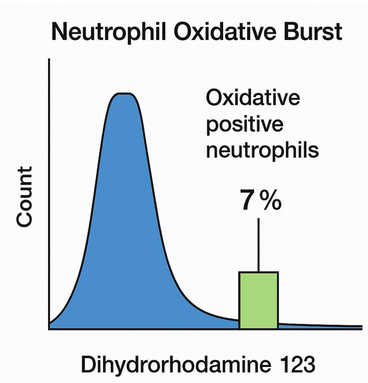
Introduction

Chronic granulomatous disease (CGD) CGD is typically diagnosed in early childhood due to its clinical severity. Delayed diagnoses in adulthood are uncommon but have been increasingly reported[1-3]. The mean age of diagnosis is 3 years for X-linked forms, and around 7 years for autosomal recessive variants, but adult-onset or delayed recognition remains a diagnostic challenge[4,5]. Here, we present a case of CGD diagnosed in the fifth decade of life.

Immunologic Evaluation

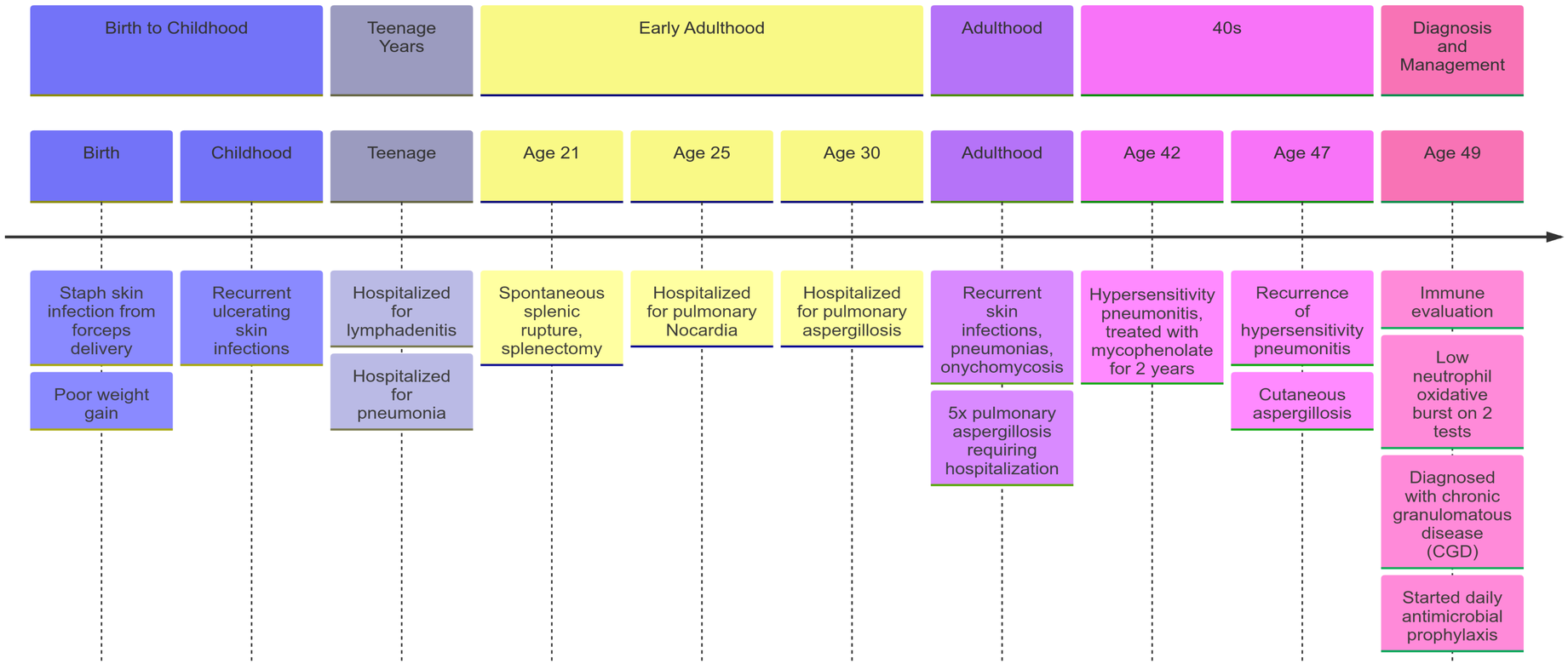
- Pneumococcal, tetanus and diphtheria titers protective
- Inborn errors of immunity genetic panel negative
- NCF1 gene sequencing pending

IgG	1290
IgA	384
IgM	61
IgE	8.7



CD3 absolute	717
CD3 %	65.9
CD3CD4 absolute	450
CD3CD4 %	41.4
CD3CD8 absolute	249
CD3CD8 %	22.8
CD4/CD8 ratio	1.8
CD16/56 absolute	230
CD16/56 %	21.1
CD19 absolute	118
CD19 %	10.8

Clinical History



Discussion

This case underscores the necessity of maintaining a high index of clinical suspicion for CGD in adult patients presenting with a typical infectious pattern. Although the patient exhibited a relatively mild phenotype during childhood, he had developed recurrent apergillosis and nocardiosis by the third decade of life, which should have prompted earlier immunologic evaluation.

Genetic analysis did not reveal pathogenic variants in known CGD-associated genes; however, many inborn error of immunity genetic testing panels do not include *NCF1* as it is a difficult gene to sequence. Mutations in *NCF1* are the most common cause of autosomal recessive CGD worldwide and are associated with higher residual NADPH oxidase activity, potentially accounting for this patient's attenuated clinical presentation and delayed diagnosis[5].

This case highlights the limitations of standard genetic panels and reinforces the importance of both comprehensive molecular diagnostics and heightened clinical awareness of CGD across all age groups.

References

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