

# Investigating Phenylalanine/Tyrosine Pathway Fluctuations in Alkaptonuria under Nitisinone Treatment

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## Abstract

### Reviewed:

**Keywords:** Alkaptonuria; Nitisinone treatment; Phenylalanine; Tyrosine pathway; Metabolite fluctuations; Enzyme activities

## Introduction

Alkaptonuria is a rare hereditary metabolic disorder characterized by the deficiency of homogentisate 1,2-dioxygenase enzyme activity, leading to the accumulation of homogentisic acid (HGA) derived from the catabolism of phenylalanine and tyrosine [1-4]. The excessive accumulation of HGA results in a range of clinical manifestations, including ochronosis, arthritis, and renal and cardiac complications. Nitisinone, a potent inhibitor of 4-hydroxyphenylpyruvate dioxygenase, has emerged as a promising therapeutic option for alkaptonuria by reducing the production of HGA. Despite its clinical efficacy, the precise impact of nitisinone on the phenylalanine/tyrosine pathway and related metabolic dynamics in alkaptonuria remains poorly understood. Understanding the alterations in the phenylalanine/tyrosine pathway induced by nitisinone is crucial for optimizing treatment strategies and elucidating the pathophysiology of alkaptonuria [5]. Therefore, this study aims to investigate the fluctuations in the phenylalanine/tyrosine pathway and associated factors in alkaptonuria patients undergoing nitisinone therapy. By elucidating the biochemical changes induced by nitisinone treatment, this research seeks to provide insights into the therapeutic mechanisms and metabolic consequences of nitisinone in alkaptonuria management.

## Materials and Methods

Study population a cohort of alkaptonuria patients undergoing

during the study period.