Ampleness of Nitisinone for the Administration of Alkaptonuria

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clinical symptomatology following initiation of nitisinone therapy. Secondary outcomes included measures of disease progression, such as musculoskeletal involvement, ochronotic pigment deposition, and renal function.

Descriptive statistics were used to summarize patient characteristics and treatment outcomes. Continuous variables were reported as mean ± standard deviation or median (interquartile range), while categorical variables were expressed as frequencies and percentages. Changes in outcome measures from baseline to follow-up were analyzed using paired t-tests or wilcoxon signed-rank tests, as appropriate [7]. is study was conducted in accordance with the principles of the declaration of helsinki and approved by the institutional review board. Informed consent was obtained from all participants or their legal guardians prior to data collection. Patient con dentiality was maintained throughout the study period. Potential limitations of this study include its retrospective design, small sample size, and potential confounding factors that could in uence treatment outcomes. Additionally, the generalizability of ndings may be limited by the single-center nature of the study and the heterogeneous nature of alkaptonuria phenotypes.

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e mean duration of nitisinone therapy was months, with a median daily dose of mg of patients experienced dose adjustments during the study period, primarily due to adherence to nitisinone therapy was high, with of patients reporting excellent compliance [8]. Following initiation of nitisinone therapy, there was a signi cant reduction in urinary homogentisic acid (HGA) levels from baseline to follow-up. e magnitude of HGA reduction varied among patients, with achieving normalization of urinary HGA levels. Improvements in clinical symptomatology were observed in of patients, with reductions in musculoskeletal pain, joint sti ness, and ochronotic pigment deposition reported. Additionally, of patients demonstrated stabilization or slowing of disease progression, as evidenced by radiographic imaging and functional assessments. However, of patients experienced persistent or progressive renal impairment despite nitisinone therapy.

e results of this study demonstrate the e cacy of nitisinone in reducing urinary HGA excretion and ameliorating clinical symptoms in patients with alkaptonuria. Consistent with previous reports, nitisinone therapy was associated with a signi cant decrease in HGA levels, indicative of decreased metabolic substrate accumulation. is biochemical response was paralleled by improvements in musculoskeletal symptoms and ochronotic pigment deposition, highlighting the potential disease-modifying e ects of nitisinone [9]. e observed variability in treatment response underscores the heterogeneous nature of alkaptonuria and the need for personalized therapeutic approaches. Factors in uencing treatment outcomes may include baseline disease severity, genotype-phenotype correlations, and individual di erences in drug metabolism and pharmacokinetics. Future studies exploring predictors of nitisinone response and optimizing treatment algorithms are warranted to maximize clinical bene t in patients with alkaptonuria. Despite the overall favorable response to nitisinone therapy [10], challenges remain in the long-term management of alkaptonuria, particularly in preserving renal function and preventing systemic complications. Strategies to mitigate renal impairment, such as early intervention with renoprotective agents and close monitoring of renal function, should be integrated into comprehensive care plans for patients with alkaptonuria. Additionally,

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