

alpha globin common mutation analysis

Alpha Globin Common Mutation Analysis: Challenges and Opportunities in Diagnosing and Treating Hemoglobinopathies

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Abstract: This article provides a comprehensive overview of alpha globin common mutation analysis, exploring the current methodologies, challenges, and future opportunities within this crucial field of medical genetics. We examine the prevalence of different alpha globin gene mutations, the diagnostic techniques employed, and the limitations faced in accurately identifying and characterizing these mutations. Furthermore, we discuss the implications of alpha globin common mutation analysis for clinical management and the potential for advancements in genetic therapies.

1. Introduction: The Significance of Alpha Globin Common Mutation Analysis

Alpha globin common mutation analysis is essential for the diagnosis and management of alpha-thalassemias, a group of inherited blood disorders affecting the production of alpha-globin chains, a key component of hemoglobin. These disorders range in severity from asymptomatic carrier states to life-threatening conditions such as hydrops fetalis. Accurate and timely alpha globin common mutation analysis is therefore crucial for appropriate genetic counseling, prenatal diagnosis, and the implementation of effective treatment strategies.

1. Common Alpha Globin Gene Mutations and Their Phenotypes

Several common mutations in the alpha globin genes (HBA1 and HBA2) are associated with alpha-thalassemia. These include deletions (e.g., $-\alpha 3.7$, $-\alpha 4.2$), non-deletional mutations (e.g., single nucleotide polymorphisms leading to missense or nonsense

mutations), and other structural variations. The phenotypic expression of alpha-thalassemia is highly dependent on the number of affected alpha-globin genes. Homozygous deletion of all four alpha-globin genes (-α/-α) results in hydrops fetalis, a severe condition usually fatal in utero. Other genotypes, such as -α/αα, -α/-α^{3.7}, or those with non-deletional mutations, can manifest as milder forms of alpha-thalassemia with variable clinical presentations. Understanding the specific genotype through alpha globin common mutation analysis is essential for predicting the severity of the disease.

1. Methodologies for Alpha Globin Common Mutation Analysis

Several techniques are currently employed for alpha globin common mutation analysis. These include:

Gap-PCR: A polymerase chain reaction (PCR)-based method used to detect deletions in the alpha-globin genes. This method is relatively inexpensive and widely accessible, making it suitable for large-scale screening programs.

Real-time PCR: A quantitative PCR technique offering high sensitivity and specificity for detecting both deletions and non-deletional mutations. It allows for the precise quantification of alpha-globin gene dosage.

Restriction fragment length polymorphism (RFLP) analysis: This technique exploits variations in DNA sequences recognized by restriction enzymes to differentiate between normal and mutant alleles. While less common now, it remains useful for detecting specific known mutations.

DNA sequencing: Provides the most comprehensive analysis, identifying both known and novel mutations in the alpha-globin genes. Next-generation sequencing (NGS) technologies have significantly increased the throughput and cost-effectiveness of this method, enabling the simultaneous analysis of multiple genes.

Multiplex ligation-dependent probe amplification (MLPA): A relatively new technique that allows for the simultaneous detection of multiple deletions and duplications within the alpha-globin gene cluster, offering high throughput and improved sensitivity compared to traditional methods.

1. Challenges in Alpha Globin Common Mutation Analysis

Despite significant advancements, several challenges remain in alpha globin common mutation analysis:

Genetic heterogeneity: The wide range of mutations in the alpha-globin genes necessitates the use of multiple diagnostic techniques to achieve comprehensive coverage.

Detection of rare and novel mutations: Many rare and novel mutations may not be detected by commonly used methods, requiring advanced techniques like NGS for complete characterization.

Interpreting complex genotypes: The presence of multiple mutations or structural variations can complicate genotype-phenotype correlations, requiring careful interpretation of analytical results in conjunction with clinical data.

Cost and accessibility: Advanced techniques like NGS can be expensive, limiting their accessibility in resource-constrained settings.

Standardization and validation: Lack of standardization in diagnostic methods across different laboratories can lead to inconsistencies in results.

1. Opportunities and Future Directions in Alpha Globin Common Mutation Analysis

Despite the challenges, several opportunities exist for improving alpha globin common mutation analysis:

Development of high-throughput, cost-effective methods: Further advancements in NGS and other molecular techniques promise to make comprehensive alpha-globin gene analysis more accessible and affordable.

Improved genotype-phenotype correlations: Large-scale studies incorporating detailed clinical data and genetic information will improve our ability to predict disease severity based on genotype.

Development of targeted therapies: A deeper understanding of the molecular mechanisms underlying alpha-thalassemia will facilitate the development of targeted therapies, such as gene editing or gene therapy.

Improved genetic counseling: Accurate and timely alpha globin common mutation analysis is critical for providing appropriate genetic counseling to families affected by alpha-thalassemia.

Prenatal diagnosis: Accurate and early detection of alpha-thalassemia through prenatal diagnosis allows for informed decision-making during pregnancy.

1. Conclusion

Alpha globin common mutation analysis is a crucial component of the diagnosis, management, and future treatment of alpha-thalassemias. While challenges remain in terms of genetic heterogeneity, cost, and accessibility, advancements in molecular diagnostic techniques and our understanding of genotype-phenotype correlations are paving the way for more accurate, comprehensive, and accessible testing. This, in turn, will improve genetic counseling, prenatal diagnosis, and the development of effective therapies for individuals affected by these inherited blood disorders.

FAQs:

1. What is the difference between alpha and beta thalassemia? Alpha thalassemia affects the alpha-globin gene, while beta thalassemia affects the beta-globin gene. Both lead to reduced hemoglobin production but have different clinical presentations.

1. How is alpha thalassemia inherited? Alpha thalassemia is inherited in an autosomal recessive pattern. Both parents must carry at least one affected alpha-globin gene to have a child with the condition.
1. What are the symptoms of alpha thalassemia? Symptoms vary widely depending on the severity of the condition. They can range from asymptomatic to severe anemia, jaundice, and splenomegaly.
1. How is alpha thalassemia diagnosed? Diagnosis involves complete blood count, hemoglobin electrophoresis, and alpha globin common mutation analysis.
1. What are the treatment options for alpha thalassemia? Treatment options vary depending on the severity of the condition and may include blood transfusions, iron chelation therapy, or in some cases, bone marrow transplantation.
1. Can alpha thalassemia be prevented? Genetic counseling and prenatal diagnosis can help prevent the transmission of alpha thalassemia to future generations.
1. What is the prognosis for alpha thalassemia? The prognosis varies greatly depending on the severity of the disease. Severe forms (hydrops fetalis) are usually fatal, while milder forms can be managed effectively with appropriate medical care.
1. What is the role of genetic counseling in alpha thalassemia? Genetic counseling provides individuals and families with information about the inheritance, diagnosis, and management of alpha thalassemia, helping them make informed decisions about family planning and healthcare.
1. What are the ethical considerations related to alpha thalassemia testing and genetic counseling? Ethical considerations include informed consent, privacy, and the potential for discrimination based on genetic information.

Related Articles:

1. "Advances in Alpha-Thalassemia Diagnosis: A Comprehensive Review of Molecular Techniques": This article would provide a detailed comparison and analysis of various molecular techniques currently utilized for alpha thalassemia diagnosis, highlighting their strengths, limitations, and suitability for different clinical scenarios.
1. "Genotype-Phenotype Correlation in Alpha-Thalassemia: A Global Perspective": This article would focus on the relationship between genetic variations and clinical manifestations of alpha-thalassemia across diverse populations, discussing the influence of genetic modifiers and environmental factors.
1. "The Role of Next-Generation Sequencing in Alpha-Thalassemia Diagnostics and Carrier Screening": This article would discuss the application of NGS technologies in alpha thalassemia diagnostics, covering its advantages in detecting novel mutations and improving the efficiency of large-scale screening programs.
1. "Prenatal Diagnosis and Management of Alpha-Thalassemia: A Multidisciplinary Approach": This article would focus on the role of prenatal diagnosis in detecting alpha-thalassemia during pregnancy, highlighting various strategies for managing affected pregnancies.
1. "Novel Therapeutic Strategies for Alpha-Thalassemia: Gene Editing and Gene Therapy": This article would explore promising new therapeutic approaches for alpha-thalassemia, such as CRISPR-Cas9 gene editing and gene therapy strategies.
1. "The Burden of Alpha-Thalassemia: Global Prevalence and Public Health Implications": This article would present epidemiological data on alpha-thalassemia prevalence worldwide, discussing its impact on public health and healthcare systems.
1. "Clinical Management of Severe Alpha-Thalassemia: Current Guidelines and Future

Perspectives": This article would provide a detailed overview of current clinical guidelines and treatment protocols for severe alpha-thalassemia, while discussing potential future improvements in management.

1. "Genetic Counseling for Alpha-Thalassemia: Best Practices and Ethical Considerations": This article would provide best-practice recommendations for genetic counseling, addressing ethical issues related to informed consent and non-directive counseling.

1. "The Impact of Alpha-Thalassemia on Quality of Life: Patient Perspectives and Psychosocial Factors": This article would focus on the psychosocial impact of alpha-thalassemia on patients and families, incorporating qualitative data obtained through patient interviews and surveys.

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common features and provide the location or mapping limits and function of responsible genes.

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2015-11-11 The book, *Inherited Hemoglobin Disorders*, describes the genetic defects of hemoglobins, disease complications, and therapeutic strategies. This book has two distinct sections. The first theme includes seven chapters devoted to the types of hemoglobinopathies, mutation spectrum, diagnostic methods, and disease complications, and the second theme includes three chapters focusing on various treatment strategies. The content of the chapters presented in the book is guided by the knowledge and experience of the contributing authors. This book serves as an important resource and review to the researchers in the field of hemoglobinopathies.

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the reader's PC or PDA.--Page 4 of cover.

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Methods and Protocols, will be introduced with a review of the great milestones in the field, and the scientists responsible for those achievements. The history of hemoglobin can be divided into three periods: the Classical period, the Modern period, and the Post-Modern period. I am inclined to include as the four major members of the classical period Francis Roughton, Quentin Gibson, Jeffries Wyman, and Linus Pauling, not only because of their achievements, but also because of the superb scientists they trained and/or influenced. Francis John Worsely Roughton (1899–1972) (Fig. 1), in his laboratory at Trinity College in Cambridge, England, made the first measurements of the rapid reaction of oxygen with hemoglobin at the millisecond scale, at first by flow-mixing methods and later by flash photolysis. He not only opened an era of molecular research of hemoglobin, but also invented the methodology for fast reactions through the use of laser technology, which was later improved by others so that even faster reactions could be detected. Another contribution of Roughton was the education of Quentin H. Gibson (Fig. 2), his favorite student, who, in his laboratory in Sheffield, continued to expand the horizon of ligand binding to hemoglobin, defining the oxygen binding constants for each of the hemes of hemoglobin. Though this did not, as expected, solve the underlying mechanism of ligand cooperativity as discussed below, it was nonetheless an important milestone.

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to accurately identify cells, simplifies hemostasis and thrombosis concepts, and covers normal hematopoiesis through diseases of erythroid, myeloid, lymphoid, and megakaryocytic origins. Easy to follow and understand, this book also covers key topics including: working in a hematology lab; complementary testing areas such as flow cytometry, cytogenetics, and molecular diagnostics; the parts and functions of the cell; and laboratory testing of blood cells and body fluid cells. - UPDATED nearly 700 full-color illustrations and photomicrographs make it easier for you to visualize hematology concepts and show what you'll encounter in the lab, with images appearing near their mentions in the text to minimize flipping pages back and forth. - UPDATED content throughout text reflects latest information on hematology. - Instructions for lab procedures include sources of possible errors along with comments. - Hematology instruments are described, compared, and contrasted. - Case studies in each chapter provide opportunities to apply hematology concepts to real-life scenarios. - Hematology/hemostasis reference ranges are listed on the inside front and back covers for quick reference. - A bulleted summary makes it easy for you to review the important points in every chapter. - Learning objectives begin each chapter and indicate what you should achieve, with review questions appearing at the end. - A glossary of key terms makes it easy to find and learn definitions. - NEW! Additional content on cell structure and receptors helps you learn to identify these organisms. - NEW! New chapter on Introduction to Hematology Malignancies provides and overview of diagnostic technology and techniques used in the lab.

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alpha globin common mutation analysis: *Evidence-Based Management of Sickle Cell Disease* M D George R Buchanan, M D M P H Araba N Afenyi-Annan, M D Samir K Ballas, 2014-09-09 Sickle cell disease can be severe and disabling. When properly treated, patients live longer and with better quality life. This is a US government publication intended to provide evidence-based guidelines for the care of these patients for the use of all concerned providers as well as patients and family members. This book is available in print here for convenience.

alpha globin common mutation analysis: *Iron Chelation Therapy* Chaim Hershko, 2002

Within the last few years, iron research has yielded exciting new insights into the understanding of normal iron homeostasis. Such development, and the evolution of improved strategies of Iron Chelating Therapy require better understanding of the pathophysiology of iron toxicity and the mechanism of action of iron chelating drugs. The timeliness of the present volume is underlined by several significant developments in recent years. New insights have been gained into the molecular basis of aberrant iron handling in hereditary disorders and the pathophysiology of iron overload. This volume highlights the impact of long term Iron Chelating Therapy using deferoxamine or the new, but controversial oral iron chelator deferiprone based on experience gained by multicenter trials, with special emphasis on survival, morbidity and drug toxicity; it reviews the development of the new and improved orally effective chelators suitable for clinical use in the near future and examines novel strategies of iron chelating treatment for the control of cell proliferation in malignant disease or malaria.

alpha globin common mutation analysis: *Thalassemia* Antonio Cao, Ugo Carcassi, Peter T. Rowley, 1982

alpha globin common mutation analysis: *Community Control of Genetic and Congenital Disorders* Ala'din A. S. Alwan, B. Modell, 1997 A comprehensive guide to recommended strategies for controlling genetic and congenital disorders in Eastern Mediterranean countries. Addressed to policy-makers and service providers, the book aims to heighten awareness of these disorders while also increasing understanding of the interventions available for prevention and control. Although all major hereditary disorders are covered, particular attention is given to haemoglobin disorders, including thalassaemia and sickle cell disease, which are extremely common in these countries. The book has fourteen chapters presented in four parts. Part one introduces the growing problem of genetic disorders, outlines options for treatment and prevention, and explains the principals of diagnosis. Current and future applications of genetic technology are also briefly discussed. Chapters in part two assess the epidemiological situation in Eastern Mediterranean countries for congenital malformation and chromosomal disorders, single gene disorders, and several common diseases, such as hypertension, diabetes, coronary heart disease, and some cancers that are now know to have a hereditary component. The genetic implications of consanguineous marriage, which remains common in some of these countries, are considered in particular detail. Available approaches for prevention are described in part three. Separate chapters cover primary health care approaches, the use of family history and population screening to identify couples or families at risk, and the roles of counseling, prenatal diagnosis, and neonatal screening. Part four offers practical advice on policies and services for prevention.

alpha globin common mutation analysis: *Color Atlas of Clinical Hematology* A. V. Hoffbrand, John E. Pettit, 2000 This highly revised and updated atlas is the third edition of an essential reference in the field of hematology. Highly illustrated, this colorful visual diagnostic aid condenses the pathogenesis, clinical, morphological, and investigative aspects of the whole range of blood disorders into one, comprehensive resource. The detailed reproduction of photomicrographs, along with concise, clinically-directed text facilitates the recognition of morphological abnormalities in blood and bone marrow cells. The highly accessible format of this text enables quick and easy reference for anyone using it. Comprehensive coverage the whole range of blood disorders is discussed Highly Illustrated in full colour aids recognition of signs in the cells Concise text with explanatory artworks enables quick and easy reference in the laboratory Explanatory artworks and tables clarifies complex principles, helpful for trainees Brings together pathogenesis, clinical, morphological, investigational and therapeutic aspects Additional coverage of: Newly discovered cytokines (interleukins 9 to 14) Molecular defects in inherited and acquired blood disorders New leukaemia types and thenew classifications of the lymphom

alpha globin common mutation analysis: *Human Hemoglobins* Howard Franklin Bunn, Bernard G. Forget, Helen M. Ranney, 1977 Since the dawn of the era of molecular biology, hemoglobin has been subjected to more scrutiny than any other protein, and Bunn, Forget, and

alpha globin common mutation analysis: *Textbook of Pediatric Hematology & Hemato-Oncology* MR Lokeshwar, 2015-08-31 Textbook of Pediatric Hematology & Hemato-Oncology is a highly illustrated, comprehensive guide to diseases and cancers of the blood in paediatrics. Fifty chapters by internationally recognised paediatric haematologists and haemato-oncologists are grouped into seven sections, covering physiology, neonatal haematology, red blood cell and white blood cell disorders, bleeding disorders, transfusion medicine, haemato-oncology and other general topics. The haemato-oncology section covers cancers of the blood and includes lymphoblastic leukaemia, myeloid leukaemia (chronic and acute), juvenile myelomonocytic leukaemia, and bone marrow transplantation. The final 'general' section features discussion on gene therapy, monoclonal antibodies and biological response modifiers. Each chapter is peer-reviewed, with a list of references ensuring authoritative content, and the book is further enhanced by nearly 200 images and illustrations. Textbook of Pediatric Hematology & Hemato-Oncology is an indispensable resource for paediatricians, practitioners and trainees. Key Points Comprehensive guide to paediatric diseases and cancers of the blood 50 peer reviewed chapters with references 198 images and illustrations

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Alpha “ ” ...

BSC CON 2.15 Genetic Testing: Hematologic Conditions (Non ...
Analysis Prothrombin (Factor II) 20210G>A Mutation Analysis (Quest Diagnostics) 81240 ;

Hemoglobinopathies HBA1/HBA2. and/or HBB Variant Analysis Alpha Thalassemia Panel ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257 Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364 D57, D56.1, D64.9 Beta Globin Gene Dosage ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257 Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364 D57, D56.1, D64.9 Beta Globin Gene Dosage ...

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CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

Concert Genetic Testing: Prenatal and Preconception Carrier ...

Analysis Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257*, 81258* O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

OVERVIEW POLICY REFERENCE TABLE - Louisiana Department ...

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257* Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364* D57, D56.1, D64.9 Beta Globin Gene Dosage ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

Hemoglobin Reims—a rare alpha globin chain variant and its ...

Reims, a rare alpha globin chain variant that eluted in HbS window. Hb analysis was performed by HPLC. Covalent reverse dot blot and refractory mutation system (ARMS) were used for ...

Concert Genetics Genetic Testing - Hematologic Conditions

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257 ; Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364 : D57, D56.1, D64.9 Beta Globin Gene Dosage ...

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Concert Genetics Genetic Testing - Prenatal and ... - Ambetter

Analysis Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 Targeted Variant - Single Test (GeneDx) HBA2 Targeted Variant - ...

Concert Genetics Genetic Testing - Hematologic Conditions

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257 ; Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364 : D57, D56.1, D64.9 Beta Globin Gene Dosage ...

Alpha Globin (HBA1 and HBA2) Deletion/Duplication

detected by deletion/duplication analysis of the alpha globin gene cluster and its HS-40 regulatory region. This individual is predicted to have a single functional alpha globin gene on both ...

Concert Genetic Testing - Prenatal and Preconception Carrier ...

Analysis ; Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 ...

Genetic Testing: Prenatal and Preconception Carrier BSC CON ...

Analysis, or Mutation Panel . Cystic Fibrosis Complete Rare Variant Analysis, Entire Gene Sequence (Quest Diagnostics) 81223 : ... HBA1, HBA2, or HBB Targeted Variant Analysis ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

Genetic Testing: Prenatal and Preconception Carrier BSC CON ...

May 1, 2024 · Analysis, or Mutation Panel . Cystic Fibrosis Complete Rare Variant Analysis, Entire Gene Sequence (Quest Diagnostics) 81223 : ... HBA1, HBA2, or HBB Targeted Variant ...

Concert Genetic Testing - Prenatal and Preconception Carrier ...

Analysis ; Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 ...

GENETIC TESTING: PRENATAL AND PRECONCEPTION ...

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 Targeted Variant - Single Test ... (GeneDx) 81361, 81362 HBA1, HBA2, or ...

Concert Genetic Testing - Prenatal and Preconception Carrier ...

Analysis ; Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

QHerit Expanded Carrier Screen - ReachMD

Jun 6, 2017 · Alpha-Globin Common Mutation Analysis (11175) a ACOG (per ancestry)7,9 Mediterranean, Middle Eastern, Southeast Asian, African, Chinese, Asian Indian Up to 94 ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

Effective: 7/1/2023 V2.2023 Last Review: 3/1/2023 GENETIC

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 5 HBA1 Targeted Variant-Single Test (GeneDx) HBA2 Targeted Variant-Single Test ...

Concert Genetic Testing: Prenatal and Preconception Carrier ...

Analysis Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 : O09, Z13, Z31, Z34, Z36, Z84 : 3 : HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) ...

CONCERT GENETIC TESTING: HEMATOLOGIC CONDITIONS ...

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257 Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364 D57, D56.1, D64.9 Beta Globin Gene Dosage ...

Laboratory Diagnosis of Hemoglobinopathies and Thalassemia

Hemoglobin (Heme+Globin) • Hemoglobin is a tetramer composed of 4 globin molecules;

2 alpha globins and 2 beta globins or beta like globins • The alpha globin chain is composed of 141 ...

Prenatal Screening for Hemoglobinopathies

Genetic Analysis 81364 • Beta-globin complete gene sequencing 81257 • Alpha-globin common deletions/mutations analysis • If negative, consider alpha-globin gene sequencing ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

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Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

Comprehensive Hemoglobin Analysis - Cincinnati Children's ...

G-gamma-globin gene G γ HBG2 Alpha-like globin genes (16p13) Alpha-1-globin gene α 1 HBA1 Alpha-2-globin gene α 2 HBA2 Disorders of Hb can be classified as qualitative or quantitative ...

Concert Genetics Genetic Testing: Hematologic Conditions

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257 ; Beta Globin (HBB) Sequencing (ARUP Laboratories) 81364 ; D57, D56.1, D64.9 : Beta Globin Gene Dosage ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

BSC CON 2.07 Genetic Testing: Prenatal and Preconception ...

Feb 1, 2024 · Alpha-Globin Common Mutation Analysis (Quest Diagnostics) HBA1 Targeted Variant-Single Test 81257, 81258 (GeneDx) HBA2 Targeted Variant-Single Test ... HBA1, ...

Concert Genetics Genetic Testing - Prenatal and ... - Ambetter

Analysis Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 009, Z13, Z31, Z34, Z36, Z84 3 HBA1 Targeted Variant - Single Test (GeneDx) HBA2 Targeted Variant - ...

BSC CON 2.07 Genetic Testing: Prenatal and Preconception ...

Alpha-Globin Common Mutation Analysis (Quest Diagnostics) HBA1 Targeted Variant-Single Test 81257, 81258 (GeneDx) HBA2 Targeted Variant-Single Test ... HBA1, HBA2, or HBB ...

Concert Genetic Testing - Prenatal and Preconception Carrier ...

Analysis ; Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 ...

BSC CON 2.15 Genetic Testing: Hematologic Conditions (Non ...

Analysis Prothrombin (Factor II) 20210G>A Mutation Analysis (Quest Diagnostics) 81240 ; Hemoglobinopathies HBA1/HBA2. and/or HBB Variant Analysis Alpha Thalassemia Panel ...

Concert Genetic Testing - Prenatal and Preconception Carrier ...

Analysis ; Alpha-Globin Common Mutation Analysis (Quest Diagnostics) 81257, 81258 O09, Z13, Z31, Z34, Z36, Z84 3 HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

LABORATORY REPORT WX00000000002353 W W

Alpha-Globin Common Mutation Analysis See Below QCRL RESULT: NO MUTATION DETECTED Interpretation: DNA testing indicates that this patient is negative for the seven ...

Concert Genetic Testing - Hematologic Conditions (Non ...

HBB Variant Analysis Alpha Thalassemia Panel (Prevention Genetics, part of Exact Sciences) 81259, 81269 D56.0, D56.9, D53.9, R70.1, D56.3, D56.8, Z86.2 2, 3, 4, 6 Alpha-Globin ...

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