

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.

2. 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 ($-\alpha/\alpha$) results in hydrops fetalis, a severe condition usually fatal in utero. Other genotypes, such as $-\alpha/\alpha\alpha$, $-\alpha/\alpha^{3.7}$, 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.

3. 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.

4. 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.

5. 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.

6. 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.
2. 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.
3. 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.
4. How is alpha thalassemia diagnosed? Diagnosis involves complete blood count, hemoglobin electrophoresis, and alpha globin common mutation analysis.
5. 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.
6. Can alpha thalassemia be prevented? Genetic counseling and prenatal diagnosis can help prevent the transmission of alpha thalassemia to future generations.
7. 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.
8. 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.
9. 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.
2. "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.
3. "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.
4. "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.
5. "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.
6. "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.
7. "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.
8. "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.
9. "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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laboratory or clinical setting, particularly in areas where thalassaemia is common either in the native population or in immigrant communities. The fourth edition has been both updated and re-organized. Three new chapters have been added on the link between alpha-thalassaemia and mental retardation, on avoidance and population control and on global epidemiology. Considerable emphasis is placed on molecular pathology reflecting the huge burst of information to have come out of this field in the last few years.

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Isam Jaber Al-Zwaini, 2018-07-11 Thalassemia is a very common disease first described by pediatrician Thomas Benton Cooley in 1925 who described it in a patient of Italian origin. At that time, it was designated as Cooley's anemia. George Hoyt Whipple, a Nobel prize winner, and W. L. Bradford, a professor of pediatrics at the University of Rochester, coined the term thalassemia in 1936, which in Greek means anemia of the sea (Thalassa means sea, and emia means blood), due to the fact that it is very common in the area of the Mediterranean Sea. This name is actually misleading because it can occur everywhere in the world. Thalassemia is not a single disease; it is rather a group of hereditary disorders of the production of globulin chain of the hemoglobin. Throughout the world, thalassemia affects approximately 4.4 of every 10,000 live births. It represents a major social and emotional impact on the patient and his family and a major burden on health services where the prevalence is high.

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colour pages in which each variant haemoglobin or combination of haemoglobins is illustrated by cellulose acetate electrophoresis at alkaline pH, agarose gel electrophoresis at acid pH, isoelectric focusing and one or more HPLC traces. For ease of reference, the atlas pages are arranged according to the retention time of each haemoglobin on HPLC, this becoming increasingly the primary technique employed in haemoglobin identification. Bringing a mix of necessary scientific expertise and clinical knowledge, each author has more than 30 years experience in the diagnosis of variant haemoglobins. Providing otherwise unavailable information, this unique and practical guide is illustrated with over 700 high quality colour digital images plus flow charts and line diagrams covers common and important haemoglobin variants, in addition to many rarer ones is an essential reference source for diagnosis in the haematology laboratory A remarkably useful book, Variant Haemoglobins will be valuable for haematopathologists, clinical and laboratory haematologists in practice and in training and all laboratory staff involved in haemoglobinopathy diagnosis.

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disorders. Written in a concise and approachable format, the book includes an overview of clinical and laboratory features of these disorders. The author focuses on the selection, performance, and interpretation of the tests that are offered by the majority of diagnostic laboratories. The book also explains when more specialist tests are required and explores what specialist referral centres will accomplish. The information on diagnosis is set in a clinical context. The third edition is written by a leading haematologist with a reputation for educational excellence. Designed as a practical resource, the book is filled with illustrative examples and helpful questions that can aid in the retention of the material presented. Additionally, the author includes information on the most recent advances in the field. This important text: • Contains a practical, highly illustrated, approach to the laboratory diagnosis of haemoglobin disorders • Includes “test-yourself” questions and provides an indispensable tool for learning and teaching • Presents new material on antenatal screening/prenatal diagnostic services • Offers myriad self-assessment case studies that are ideal for the trainee Written for trainees and residents in haematology, practicing haematologists, and laboratory scientists, Haemoglobinopathy Diagnosis is an essential reference and learning tool that provides a clear basis for understanding the diagnosis of haemoglobin disorders.

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allogeneic HSCT is a curative option, it is limited by the availability of matched donors, which are often available only to 15-20% of patients. An alternative to allogeneic HSCT is genetic correction of autologous HSCs, to overcome donor availability & immune side effects. This Book reviews the progress made on additive gene therapy approaches & the current state of the field. Finally, targeted genetic correction is emerging as a novel therapeutic strategy in the hemoglobinopathies. Although ideal, the inefficiency of targeted correction was rate limiting for translation of this technology to the clinic. With advancements in zinc finger nucleases and TALE endonuclease mediated targeted correction, correction frequencies in hematopoietic stem cells is now reaching levels that may become clinically relevant. Furthermore, the ability to generate autologous embryonic stem cell like cells from primary somatic cells (skin fibroblasts or hematopoietic cells) of the affected individual has allowed for the potential application of genetic correction strategies. This Book reviews upcoming genetic strategies to reactivate fetal hemoglobin production and research advances.

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Kuliev, Svetlana Rechitsky, Oleg Verlinsky, 2014-04-22 Based on one leading center's experience with over 100,000 cases, the new edition of this extensively illustrated atlas provides a detailed manual for procedures and techniques in preimplantation genetic diagnosis. New topics in this edition include de novo mutations, diseases with genetic predisposition, and HLA typing. The book provides insight f

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hemoglobin was the crucial advance that gave birth to the scientific discipline of human molecular genetics. In subsequent years, with the continued expansion of knowledge about the biology and genetics of the hemoglobins, and particularly as a result of the characterization of the very large numbers of globin gene mutations, the human hemoglobin system has remained as the premier model of gene expression at the molecular level in man. With the recent explosion of new information about the genetic properties of the hemoglobins, it appears inevitable that this gene system will continue to occupy a unique position in human molecular genetics for many years in the future. Hemoglobin genetics has also recently come of age as a diagnostic and clinical discipline. The heightening of public awareness in recent years about sickle cell disease, thalassemia, and other inherited disorders has brought increasing demands for carrier detection services as well as for genetic counseling and education. The more recent development of practical and reliable methods for the antenatal diagnosis of hemoglobin disorders has further increased the scope of clinical hemoglobin genetics, and it can be anticipated that these potent diagnostic techniques will have increasing application in the years ahead.

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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 2.15 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 ...

Alpha Globin Common Mutation Analysis Copy - x-plane.com

The Enigmatic Realm of Alpha Globin Common Mutation Analysis: Unleashing the Language is Inner Magic In a fast-paced digital era where connections and knowledge intertwine, the ...

Alpha Globin Common Mutation Analysis (2024)

Alpha Globin Common Mutation Analysis This is likewise one of the factors by obtaining the soft documents of this Alpha Globin Common Mutation Analysis by online. You might not require ...

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 ...

Alpha Globin Common Mutation Analysis Full PDF - x ...

Alpha Globin Common Mutation Analysis eBook Subscription Services Alpha Globin Common Mutation Analysis Budget-Friendly Options 6. Navigating Alpha Globin Common Mutation ...

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Alpha Globin Common Mutation Analysis eBook Subscription Services Alpha Globin Common Mutation Analysis Budget-Friendly Options 6. Navigating Alpha Globin Common Mutation ...

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 - ...

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 009, 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 Gy HBG2 Alpha-like globin genes (16p13) Alpha-1-globin gene $\alpha 1$ HBA1 Alpha-2-globin gene $\alpha 2$ 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 O09, 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 ...