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ETIOLOGICAL AND PATHOGENETIC ASPECTS OF ACROMEGALY IN PEDIATRIC PATIENTS

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Abstract: Acromegaly in children is a rare endocrine disorder characterized by excessive secretion of growth hormone (GH), usually caused by a pituitary adenoma. Unlike adults, excessive growth hormone production in children often occurs before epiphyseal plate closure, resulting in accelerated linear growth and gigantism. The disease is associated with elevated levels of insulin-like growth factor-1 (IGF-1), which mediates many of the physiological and pathological effects of growth hormone. Early diagnosis is challenging due to the gradual onset of symptoms and the rarity of the condition. Understanding the etiology and pathogenesis of pediatric acromegaly is essential for timely diagnosis, appropriate treatment, and prevention of long-term complications. This article reviews the major etiological factors and pathogenic mechanisms involved in the development of acromegaly in children.

Keywords: acromegaly, pediatric acromegaly, gigantism, growth hormone, insulin-like growth factor-1, pituitary adenoma, endocrine disorders, etiology, pathogenesis, children.

Introduction

Acromegaly is a chronic endocrine disorder resulting from prolonged hypersecretion of growth hormone (GH) and increased production of insulin-like growth factor-1 (IGF-1). In pediatric patients, excessive secretion of growth hormone before the closure of the epiphyseal growth plates leads primarily to gigantism, characterized by abnormal linear growth and excessive height. Although acromegaly and gigantism share similar hormonal mechanisms, gigantism is the predominant manifestation in children and adolescents. Pediatric acromegaly is an uncommon condition, with the majority of cases resulting from growth hormone-secreting pituitary adenomas. In rare instances, genetic syndromes such as Multiple Endocrine Neoplasia Type 1 (MEN1), McCune-Albright syndrome, Carney complex, and Familial Isolated Pituitary Adenoma (FIPA) may contribute to disease development. These genetic abnormalities affect cellular signaling pathways and endocrine regulation, leading to excessive growth hormone production. The pathogenesis of acromegaly involves complex interactions between the hypothalamus, pituitary gland, and peripheral tissues. Excessive growth hormone stimulates the liver and other tissues to produce IGF-1, which promotes cellular proliferation, skeletal growth, and soft tissue enlargement. Persistent elevation of GH and IGF-1 levels results in characteristic clinical manifestations, including accelerated growth, enlargement of the hands and feet, facial bone overgrowth, metabolic disturbances, and cardiovascular complications. Due to its rarity and progressive nature, pediatric acromegaly often remains undiagnosed for extended periods. Early recognition of etiological factors and pathogenic mechanisms is essential for improving clinical outcomes and preventing irreversible complications. Therefore, a comprehensive understanding of the disease is crucial for healthcare professionals involved in pediatric endocrinology and child healthcare.

Materials and Methods

This study was conducted through a comprehensive review of scientific literature concerning the etiology and pathogenesis of acromegaly in children. Relevant data were collected from peer-reviewed medical journals, endocrinology textbooks, clinical guidelines, and publications issued by international healthcare organizations. The literature search focused on studies investigating growth hormone (GH) hypersecretion, insulin-like growth factor-1 (IGF-1) activity, pituitary adenomas, genetic predisposition, and molecular mechanisms involved in pediatric acromegaly. Sources published within the last two decades were prioritized to ensure the inclusion of current scientific evidence. The collected information was systematically analyzed and categorized according to etiological factors, hormonal disturbances, genetic influences, and pathogenic pathways. Comparative analysis was used to identify common mechanisms contributing to disease development and progression. The findings were synthesized to provide a comprehensive overview of the causes and pathophysiological processes underlying acromegaly in children.

Results

The analysis of the reviewed literature demonstrated that growth hormone-secreting pituitary adenomas are the primary etiological factor responsible for most cases of acromegaly in children. These adenomas originate from somatotroph cells of the anterior pituitary gland and lead to uncontrolled secretion of growth hormone. Several genetic disorders were identified as important contributors to disease development. Conditions such as Multiple Endocrine Neoplasia Type 1 (MEN1), Familial Isolated Pituitary Adenoma (FIPA), Carney Complex, and McCune-Albright Syndrome were associated with an increased risk of pituitary tumor formation and excessive hormone production. The reviewed studies revealed that elevated growth hormone levels stimulate the liver and peripheral tissues to produce increased amounts of insulin-like growth factor-1 (IGF-1). High concentrations of IGF-1 promote excessive bone growth, soft tissue proliferation, and metabolic alterations. In children, these hormonal changes result in accelerated linear growth, abnormal height increase, enlargement of extremities, and characteristic facial changes. Additional findings indicated that prolonged exposure to elevated GH and IGF-1 levels may lead to cardiovascular abnormalities, insulin resistance, glucose intolerance, headaches, visual disturbances, and musculoskeletal complications. The severity of clinical manifestations was found to correlate with the duration of hormone excess and tumor size.

Discussion

The findings of this review confirm that pediatric acromegaly is a rare but clinically significant endocrine disorder characterized by excessive secretion of growth hormone and insulin-like growth factor-1. The disease primarily develops as a result of pituitary adenomas, although hereditary and genetic factors may also play an important role. The pathogenesis of acromegaly involves dysregulation of the hypothalamic-pituitary axis. Excessive secretion of growth hormone stimulates continuous production of IGF-1, which acts as the principal mediator of abnormal tissue growth. Unlike adults, children experience accelerated skeletal growth because their epiphyseal growth plates remain open. Consequently, gigantism frequently develops before the characteristic manifestations of acromegaly become evident. The reviewed literature emphasizes the importance of genetic factors in pediatric cases. Mutations affecting cellular proliferation and endocrine regulation contribute to tumor formation and hormonal imbalance. Understanding these mechanisms is essential for improving diagnostic accuracy and developing targeted therapeutic strategies. Furthermore, delayed diagnosis remains a major clinical challenge because the disease progresses gradually and early symptoms may be overlooked. Early identification of abnormal growth patterns, hormonal abnormalities, and pituitary tumors is crucial for preventing irreversible complications. Modern treatment approaches, including surgical intervention, medical therapy, and

radiotherapy, have significantly improved patient outcomes when initiated promptly. Overall, a thorough understanding of the etiological and pathogenic mechanisms of acromegaly in children is essential for early diagnosis, effective treatment, and long-term management of affected patients.

Conclusion

Pediatric acromegaly is a rare endocrine disorder primarily caused by excessive secretion of growth hormone, most commonly due to pituitary adenomas. Genetic syndromes such as MEN1, Carney complex, McCune-Albright syndrome, and familial isolated pituitary adenoma also contribute to disease development. The pathogenesis is characterized by dysregulation of the hypothalamic–pituitary axis, leading to increased production of growth hormone and insulin-like growth factor-1 (IGF-1), which are responsible for abnormal skeletal growth and metabolic disturbances. In children, the presence of open epiphyseal growth plates results in gigantism rather than classical adult acromegaly features. Delayed diagnosis is common due to the gradual onset of symptoms, which can lead to severe complications involving the cardiovascular, metabolic, and musculoskeletal systems. Early recognition of abnormal growth patterns, hormonal imbalance, and neuroimaging findings is essential for timely diagnosis and management. A multidisciplinary approach involving endocrinologists, neurosurgeons, and pediatric specialists is crucial for improving patient outcomes. Overall, understanding the etiological and pathogenic mechanisms of pediatric acromegaly is fundamental for early intervention and prevention of long-term complications.

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