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Abstract

Role of Brachytherapy in the Treatment of Endocavitary Lesions Associated With Costello Syndrome

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Abstract:

Costello syndrome is a rare genetic disease first described in 1971 by New Zealand doctor Jack Costello, from whom the name derives. 14 years after the first report, its actual existence was confirmed by two American authors, who expanded its distinctive characteristics. A true incidence of Costello syndrome has not been calculated. Over 100 cases of affected patients are described in the literature.

In assisting patients affected by this syndrome, the collaboration of various specialists such as the pediatrician-dysmorphologist, the cardiologist, the endocrinologist, the child neuropsychiatrist, the orthopedist, the physiatrist, the ophthalmologist, the otolaryngologist, etc. is necessary. It is important that the management of such patients is entrusted to doctors who know the problems of the syndrome and to structures suited to their needs in order to be able to intervene favorably and with adequate therapeutic and rehabilitation measures.

It is due to modification of the genetic material, which cannot be detected with the standard karyotype examination. Most of the known cases have been described in families with healthy parents and siblings, thus suggesting that the origin of the syndrome is the de novo alteration of a single gene (autosomal dominant de novo mutation). Recently, the mutation of a gene located on chromosome 11, the HRAS gene, which is part of the RAS protooncogene family, has been identified in approximately 80% of patients.

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The characteristic elements of the syndrome are represented by growth retardation, psychomotor retardation, facial anomalies, skin anomalies, heart disease, skeletal and joint alterations. The complete clinical picture is not present from birth, the clinical phenotype evolves over time and some clinical signs appear later. In a variable percentage, children affected by Costello syndrome may present other clinical disorders such as visual disorders (strabismus, refractive errors), scoliosis, sleep disorders (in particular apnea), tendency to the development of tumors (neuroblastoma, rhabdomyosarcoma, bladder tumors).

Diagnosis

The diagnosis is essentially clinical. Some phenotypic elements and the natural history of the disease are quite characteristic of this condition and allow the diagnostic classification. The recent discovery of the gene allows the clinical diagnosis to be confirmed in approximately 80% of cases.

Prognosis

The pathology has a chronic course. Patients must be closely monitored and frequent medical checks are necessary. The main cause of morbidity and mortality is represented by a serious cardiac pathology or the development of tumor pathology. In the absence of this, the average survival is generally similar to that of the normal population. Role of brachytherapy in the treatment of endocavitary lesions associated with Costello syndrome. The mission is to improve the therapeutic index of pediatric tumors with radiotherapy with the aim of increasing local control of the disease in a multidisciplinary clinical management context and reducing the extent of possible medium-long term sequelae in a growing organism. Three pediatric patients (2/3/4 years) suffering from rare diseases were treated and treated with Brachytherapy through the use of specific applicators; the visits to the Pediatric RT Unit were daily and performed personally in collaboration with the IOV Head Doctor; The treatments are those foreseen by the SIOP Protocols; the treatment outcomes are monitored at 2/4/6 months to check the healing processes and the compatibility with the other treatments proposed by the Oncohematology UOC;

Instrumental diagnostic checks performed according to SIOP Protocols