International Journal of Current Advanced Research
ISSN: O: 2319-6475, ISSN: P: 2319-6505, Impact Factor: 6.614
Available Online at www.journalijcar.org
Volume 7; Issue 10(D); October 2018; Page No. 15959-15960
DOI: http://dx.doi.org/10.24327/ijcar.2018.15960.2928

Research Article

STICKLER'S SYNDROME

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ARTICLE INFO

Article History:

Received 15th July, 2018
Received in revised form 7th August, 2018
Accepted 13th September, 2018
Published online 28th October, 2018

Key words:

Ophthalmogical, vitreous gel, prophylactic peripheral retinal cryotherapy

ABSTRACT

Stickler syndrome is a genetic disorder with characteristic ophthalmological and orofacial features, deafness and arthritis. Abnormalities of vitreous gel found within the eyes. The cause of stickler's syndrome is COL2A1 and COL11A1. The estimated prevalence rate is 1-3 per 10,000 births and at 1 per 7,500 birth Signs and symptoms is eye problem hearing deficit, joint and bone deformity. The treatment of stickler syndrome is therapy like speech, physical, hearing aids and special education, preventive measures stickler syndrome is genetic counselling and prophylactic peripheral retinal cryotherapy.

INTRODUCTION

Stickler syndrome is a group of hereditary conditions characterized by a distinctive facial appearance, eye abnormalities, hearing loss and joint problems. Stickler's syndrome refers to a group of disorders of connective tissue.

Definition

Stickler's syndrome is a genetic disorder that can cause serious vision, hearing and joint problems. A characteristic feature of stickler syndrome is a somewhat flattened facial appearance.

Incidence

Prevalence rates have been estimated at 1-3 per 10,000 birth and at 1 per 7,500 birth.

Etiology

Stickler syndrome is caused by mutations in certain genes involved in the formation of collagen- one of the building blocks of many types of connective tissues. Between 80-90 % of all cases are classified as type I and are caused by mutations in the COL2A1 gene. 10-20 % of casesare classified as type II and are caused by mutation in the COL11A1 gene.

Pathophysiology

Stickler syndrome is believed to be a direct result of abnormalities in the production of collagen types II, IX and XI identical(homotrimer) or differing (hetrotrimer) polypeptide chains; genetic mutations affecting the ability of constituent polypeptide chains; genetic mutations affecting the ability of constituent polypeptide chains to successfully form stable trimer therefore prevent the production of mature collagen and subsequently produce the clinical manifestation of stickler syndrome

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Clinical Manifestations

Eye problem- Glaucoma, cataract and retinal detachment
Hearing deficit- Ability to hear high frequencies
Bone and joint abnormalities- Abnormal curvature of the spine, such as scoliosis. Osteoarthritis

Diagnostic Evaluation

Imaging tests- X-rays can reveal abnormalities or damage in the joints and spine

Eye exams- To check cataracts and glaucoma

Hearing tests- These tests measures the ability to detect different pitches and volumes of sound

Genetic testing- Genetic testing can also be used to help in family and to determine your risk of passing on the gene to your children when the hereditary pattern is not clear from the family history.

Management

Therapy

Speech therapy- Child may need speech therapy if hearing loss interferes with his or her ability to learn how to pronounce certain sounds

Physical therapy- In some cases, physical therapy may help with mobility problems associated with joint pain and stiffness. Equipment such as braces, canes and arch supports also may help

Hearing aids- Child has problems hearing, you may find that his or her quality of life is improved by wearing a hearing aid
Special education- Hearing or vision problems may cause learning difficulty in school, so special education services may be helpful

Surgery

Tracheostomy
Jaw surgery
Cleft palate repair
Eye surgeries
Ear tubes
Joint replacement
Spinal bracing or fusion surgeries

Prevention

Primary prevention-Genetic counseling

Prophylactic peripheral retinal cryotherapy may be effective in preventing development of rhegmatogenous retinal detachment.

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How to cite this article:

Hemavathy V., Binipaul V.J and Jannet Susannal (2018) 'Stickler's Syndrome', *International Journal of Current Advanced Research,* 07(10), pp. 15959-15960. DOI: http://dx.doi.org/10.24327/ijcar.2018.15960.2928

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