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Research Article

A STUDY REVIEW OF WILLIAM BUREN SYNDROME AS A MULTI SYSTEMIC DISORDER AND THE COMPLICATION WHICH ACCOMPANIES

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A B S T R A C T

Introduction: William syndrome is a congenital disease which affects multiple systems at the same time. It involves the cardiovascular system, connective tissue, central nervous system, also causes psychological and psychiatric disorders. These populations will be presented with many characteristic facial features, dermatological, gastrointestinal, urogenital and endocrine. They may face complications during anesthesia. This study is a review of previous study showing and I define which complication of each system Williams syndrome can cause.

Conclusion: The review showed the cardiovascular system id the most commonly involved and its association with anesthesia. Fallowed by the neurological, the psychiatric and psychological in the shape of neurological delay, hypotonia, anxiety disorder and hyper sociality respectively. Also cerebellum Through The vermis and the medulla (The swallowing center, the nuclei of the 10th and 9th nerves) they contributes in the swallowing mechanism and the motility of the esophagus. William syndromes individuals will be presented with cerebellar vermis hypoplasia, ventriclomegally, thin corpus callosum, white matter immaturity and posterior fossa cysts all these complications will induce a defect in the esophageal motility. A study showed that hypercalcemia is mostly developed during infantile and early childhood.

INTRODUCTION

William syndrome develops due to a is the deletion of ELN gene which encode for elastin and other 28 genes from chromosome 7 (q11.23) And they will have characteristic facial features Which 's known with "elfin face" and upon examination of those population, They will show a broad forehead, periorbital fullness, flattened nasal bridge, wide moth and full lips, high round cheeks and a pointed chin. They also have a social and cocktail personality, and an IQ of 50-60. But they have a good verbal communication skills .Combined with hyper activity so they can be diagnosed with attention deficit disorder or anxiety disorder. (1,4,10)

One of the most common system affected of WS which develops in 80% of population is evolving the cardiovascular system. Deletion of ELN gene will cause deletion of the elastin which is a primary element in the blood vessels' wall development. Elastin allows the store of energy during systole and release of it during diastole.(1)

Supravalvular aortic stenosis is the most common cardiovascular defect which covers 45-75% of the cardiovascular defects.

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Other defects includes pulmonary artery stenosis with 37-75%, Renal artery stenosis with 7-58% which is highly associated with hypertension, coronary arteries stenosis or dilatation with 9% and sudden cardiac death 1% specially preanesthetic or peri procedure. ECG is the tool used for diagnosis which shows 60% right ventricular hypertrophy.(1)

Cranial Blood vessels can also be affected. The Middle cerebral artery is the most affected with stenosis or an emboli (Thromboemboli) from the heart .Which was shown in an MRA study was done for 27 participants in 2014 as a case-control study with 14 WS and 13 control. They expressed similar patency for a brain stroke and Hemorrhagic infraction.(2)

There was also a study was done in 2009 with 29 participants and 108 anesthetics were used 11% of the anesthetic used caused complications the most common complication was bradycardia, hypotension and cardiac ischemia . For patient who already had supravalvlar aortic stenosis 100% (all of them) developed these symptoms. Halothane and suxamthonium had the most adverse effect specially bradycardia. Replacement of anesthetics was done to reduce the severity with fenytal and atracurium but symptoms also developed. The best tool was used for diagnosis was cardiac catheterization it was more effective than ECG or Echocardiogram. (7)

For their psychological status was detected that hyper-sociality and cocktail personality is a common developmental disorder. They have an overfriendly personality which increases by age. With excessive empathy and poor social judgment .It has also been found that they don't have the ability to detect social fear so they don't show enough arousal for angry faces. (3,4,10) The etiology for the delay in their neurological development is the improper development of their cortex due to a deletion of the genes responsible for development and regulation of the brain , its cells, synapses and neurotransmitters. All that will lead to social disabilities.

Combined with a dysfunction in their frontal lobe. The can show less inhibitory control and suppression for their social behavior.(3,4,10)

Neurological development abnormality as seizure was found in 18% of the population also hypotonia and abnormal gate was the most common neurological development with 88%.For psychology and psychiatry anxiety disorder was the most common of 71% (10)

Willasms syndrome can affects the skin, A study was done in number of population in 2012 for dermatological evaluation. It's known that collagen and elastin makes 81% of the of the skin and any disturbance in their ratio will lead to skin disorders. Premature hair graying was observed in 58% of the population affected with Williams syndrome. Face wrinkling was detected in 92% of the population, wrinkles was noted in a 7 years old boy. And hyperkeratosis was found in 20% of the population. Participants from the age 10-37 showed abdominal stretch marks and the patients were with normal BMI.(5)

They can also develop hypercalcemia which's more severe during infancy and then it decreases at childhood. They will be complaining of hypotonia, constipation and abdominal pain due to the high level of calcium.

It was also found that hypercalcemia is highly associated with nephrocalcinosis which was detected in 5% of the cases in un US imaging study. (6,9) Which might resolve when they reach childhood and which's found 50% of WS population. (9)

Other systems includes gastrointestinal, Endocrinology and musculoskeletal through a study which was done in 2015 for 64 participant The gastrointestinal abnormality were : chronic constipation was the most common of 81% which is related to low muscle tone, slow motility because of neurotransmitters imbalance and the neurological development.

Endocrinology abnormalities were hypothyroidism and growth hormone deficiency covered 10 % of the cases. Musculoskeletal abnormalities covered 28% of hyperflexible joint, pectus excavatum, lordosis and avascular hip necrosis.(10)

CONCLUSION

It has been found that the Williams's syndrome can affect more than one system and the study review has proven that the cardiovascular system is the most affected in the form of supravalvular stenosis which was detected through Egg by showing hypertrophy of the left ventricle in 80% of population. Which can also lead to complication during anesthesia due to the supravalular stenosis in the form of brady cardia or hypotension (1). And the fallowing system is involving the Brain wither neurodevelopmental or psychiatric or psychological. Neurodevelopmental delay and hypotonia or anxiety disorder respectively 88% of all CNS disorder wither neurological or psychological/psychiatric.(10) They can also develop hypercalcemia which's more severe during infancy and then it decreases at childhood. They will be complaining of hypotonia, constipation and abdominal pain due to the high level of calcium. The gastrointestinal abnormality were: chronic constipation was the most common of 81% which is related to low muscle tone, slow motility because of neurotransmitters imbalance and the neurological development. I have also found Achalasia can be a complication of Williams syndrome Which's associated with hypercalcemia specially in infantile, early childhood and sometimes it continues until adulthood. Hypercalcemia will induce a functional and a neuromuscular dysphagia, Due to nervous system depression. It will also reduce the contractility of the smooth muscle of the esophagus specially the lower 2/3 which 's highly related to the development of achalasia and dysphagia.(11)

Also cerebellum Through The vermis and the medulla (The swallowing center, the nuclei of the 10^{th} and 9^{th} nerves) they contributes in the swallowing mechanism and the motility of the esophagus. (12) William syndromes individuals will be presented with cerebellar vermis hypoplasia, ventriclomegally, thin corpus callosum, white matter immaturity and posterior fossa cysts all these complications will induce a defect in the esophageal motility. (12)

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