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CENTRE FOR ADVANCED PEDIATRIC AND ADULT REHABILITATION

Epidemiology Overview

WILLIAM SYNDROME

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Described by “William an Beuren” in 1961

Rare genetic Condition

The clinical manifestations include a distinct facial appearance, cardiovascular anomalies that may be present at birth or may develop later in life, idiopathic hyper calcemia and a characteristic developmental and behavioral profile.

Path Physiology:-

Haploinsufficiency due to a deletion at chromosome band 7q11.23 that involves the elastin gene (ELN) is implicated.

- **William syndrome is not solely caused by elastin haploinsufficiency; the deletion involves a region that spans more than 28 genes – Contiguous gene deletion syndrome.**
- **Copy number variants in the 7q11.23 region have been found to be associated with autism in a study of over 4000 individuals who did not have William syndrome.**

Epidemiology:-

- **US – 1/7500 -20,000 birth**
- **Mortality – Cardiovascular disease**
- **Sudden Death – SVAS, Severe pulmonary stenosis and myocardial ischemia secondary to either coronary insufficiency or biventricular outflow tract obstruction with ventricular hypertrophy.**
- **Hypertension - 50%**
- **Renal artery stenosis**
- **A higher frequency of obesity, impaired glucose tolerance and diabetes mellitus have been found in adults with William syndrome compared to the general population**

- **Increased TSH. Increased prevence in points with William syndrome.**
- **GI problems – feeding problems and colic , reflox and chronic constipation**
- **Sigmoid diverticolitis in adults more in William syndrome.**
- **It is a multisystem condition with other potential consequences including developmental delay, motor delay, hearing loss, severe dental disease, ocular problems, progressive joint contractures, nephrolithiasis, bowel and bladder diverticula.**

Gender:-M/f

- A greater severity and earlier presentation of cardiovascular disease may be observed in males.

Age:- Birth to adulthood

- Features that may be detected antenatally include the characteristics cardiovascular lesions.
- Fetal ultrasonography of neonates with William syndrome has revealed multicystic dyplastic kidney in addition to congenital heart lesions.
- Associated findings on prenatal screening that have been reported include an increased fetal nuchal transluency and low maternal serum alpha fetoprotein (MSAFP)

Clinical Presentation:-

It includes a pattern of growth and development and a specific neurodevelopment profile primarily involving 4 areas:-

1. Cognitive Development
2. Language
3. Auditory function
4. VIsuospatial function

- Children have prenatal and postnatal growth delay and usually present with failure to thrive
- Short stature
- Poor weight gain and feeding problems
- Recurrent middle ear infections.

- **Visual disturbances – mainly related to esotropia, cataracts and hyperopia in as many as 50% of individuals with William syndrome**
- **Congenital heart disease and hypertension**
- **In children – Increased urinary frequency and daytime wetting**
- **Renal abnormalities – hypercalcemia and hypercalciuria**
- **Delayed bone age and decreased insulin like growth factor levels may be noted.**
- **Early pubertal onset**
- **Glucose tolerance or overt diabetes mellitus in more than 20 years.**
- **Subclinical hypothyroidism**

- **Connective tissue abnormalities such as abnormal joint mobility, hemias and diverficula are possible.**
- **Chronic abdominal pain and they are at increased risk for cardiac disease.**
- **Mild to moderate mental retardation**
- **Early language acquisition is delayed. The quality and affect of speech are relatively normal.**
- **Visual –spatial problem impact daily life with difficulties in handwriting, drawing and gait apraxia especially on uneven or sandy surface.**

- **As many as half of all children with William syndrome may exhibit autism spectrum social and communicative deficits**
- **They are overfriendly, hyperactive, inattentive and hypersensitive to loud sounds or certain types of sounds.**
- **Adults may have a high rate of emotional and behavioral problems, poor social relationship and anxiety, preoccupations and obsessions, phobias, panic attacks and depression.**

Physical:-

- **Children with William syndrome are generally full-term infants with prenatal growth delay**
- **Microcephaly is observed in 1/3rd of children and postnatal failure to thrive is typical**

Children and adult facial features:-

Short upturned nose,flat nasl bridge, long philtrum, flat malar area, wide mouth full lips, dental malocclusions and widely spaced teeth, micrognathia and periorbital fullness.

Astellate, lacy pattern of the irises can be observed in children with blu eyes.

The voice may be hoase.

Nails tend to be hypoplastic and the skin soft and lax; the halluces have a valgus deviation.

Dental – small and peg shaped teeth, increased interdental spacing, absence of on or more primary or secondary teeth, anterior cross-bitte malocclusion and excessive gingival tissue.

Other findings:- Hyperacusis(despite hearing loss), hoarse voice, joint hyperelasticity, contractures, kyphoscoliosis and lordosis.

Generalised hypotonia is found in infants with William syndrome and progress to spasticity age.

CONCLUSIONS

If you have any questions to ask about Best Treatment for Williams Syndrome in Bangalore ! Or Are you still worried and confused about any syndrome, to find out more.

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