Extended Abstract Journal of Congenital Disorder 2019 Vol.1 Issue.2

The First Case of Cutis Laxa Type II (Debre Type) Associated with Atrial Septal Defect

Aamir J Al-Mosawi

Senior Advisor and Doctor, Children Teaching Hospital, Medical City, Israel

Abstract

Cutis laxa type II (MIM 219200) is an uncommon inherent issue, first depicted by Debre et al. in 1937. During the period from 1937 to 2008, there have been 34 (25 females and 9 guys) very much reported distributed instances of this uncommon issue.

In 2009, I announced the thirty fifth instance of this issue and I additionally looked into the recently distributed and thirty four all around recorded cases. A dysmorphic young lady was alluded at four years old years (early April 2016) on account of skin variation from the norm and deferred discourse. The young lady had cutis laxa with repetitive free skin present from birth. This skin variation from the norm was obvious on the appendages and mid-region. Dysmorphic facial component included, wide and discouraged nasal scaffold, and the eyes were wide set with downslanting palpebral crevices. Assessment of the cardiovascular framework was ordinary aside from the nearness of a systolic mumble. Echocardiography uncovered the nearness of a little atrial septal defect.In this paper, I am announcing the 36th instance of Cutis laxa (Debre type) which is the principal case to be related with atrial septal imperfection.

Cutis laxa is a heterogeneous gathering of inherent and seldom procured scatters. In the inborn kind, there is hereditary heterogeneity with proof of autosomal predominant, passive or X-connected passive legacy. The autosomal prevailing sort is because of transformation in the versatile quality with a benevolent course and an odd life expectancy. Different signs of this sort (AD) are pneumonic supply route stenosis, emphysema, bronchiectasia, hernia and genital prolapse.

The autosomal passive cutis laxa (ARCL) incorporates type I (as the threatening structure present during childbirth or soon after birth) and type II cutis laxa (wrinkly skin condition and De Barsy disorder). Type I ARCL is because of transformation of fibulin quality .This sort is portrayed by early puerile pneumonic emphysema, hernia and numerous diverticula and has poor anticipation as one of the two particular issue of intrinsic cutis laxa depicted first by Agha et al. A lack in lysyl oxydase, which is firmly identified with wrinkly skin disorder, has been proposed in type II ARCL. De Barsy condition is related with mental hindrance, short height and corneal obfuscating A "pseudoathetoid" development issue with beginning in the second year of life gives off an impression of being a striking element in this type of inborn cutis laxa .

The principal metabolic sickness revealed in relationship with wrinkled, inelastic, old looking and listing skin was Menkes malady. This inherent metabolic illness with a X-connected passive legacy shows foundational

copper lack overwhelmingly in the cerebrum .The X-connected latent (likewise called Ehlers-Danlos type IX condition or gentle Menkes disorder) has been portrayed due to ATP7A inadequacy. Dissimilar to Ehlers-Danlos disorder, the skin in cutis laxa, albeit free, isn't hyperextensible .

Procured cutis laxa grows suddenly or after a febrile ailment, provocative skin illnesses, for example, lupus erythematosus or erythema various, amyloidosis, urticaria, angioedema and touchiness responses to penicillin .An instance of a 27-year-old female patient who created extreme aspiratory and heart contribution in adulthood most likely experiencing the gained kind of cutis laxa is accounted for .In another examination, cutis laxa condition was liable for Zenker and epiphrenic diverticula .Gastrointestinal diverticulosis is an incessant finding in ARCL type I, however it is additionally found in Ehlers-Danlos disorder and by chance in Noonan condition and numerous endocrine neoplasia type IIa and IIb.

Our introduced case was a neonate with skin laxity, respiratory trouble, heart irregularities (ASD, VSD, MR, TR), bladder diverticulosis, GI diverticulosis and hypertrophic pyloric stenosis. Bladder diverticula could be essential (Hutch, Urachal or different areas), auxiliary to impediment (back urethral valve), neurogenic brokenness, iatrogenic or related with different conditions (Prune stomach illness, Williams disorder, Ehlers-Danlos, Menkes disorder and cutis laxa) .

Attendant presence of parent affiliation, early demise in influenced kin and diverticulosis infers that this case possibly an ARCL type I.

Hypertrophic pyloric stenosis is a procured issue increasingly normal in young men and most likely first - conceived babies. There are some inclining elements, for example, erythromycin utilization or delayed pylorospasm. Hereditary elements may likewise be significant since it is increasingly basic in babies with an influenced parent or kin. There are a few reports of CL convoluted with HPS like our case .To search for any relationship among CL and HPS, it is exceptionally recommended to concentrate on further CL cases entangled with HPS.

Relating creator:

References

1. Genevieve D, Baumann C, Huber C, Faivre L, Sanlaville D, Bodemer C, et al. A tale type of syndromic cutis laxa with facial dysmorphism, congenital fissure, and mental hindrance. J Med Genet. 2004;41(6):ee77.

2. Nesibe A, Fikriye S, Muhsin S, Melda Ç. Autosomal Recessive Form of Congenital Cutis Laxa: More Than the Clinical Appearance. Pediatr Dermatol. 2002;19(5):412–414. doi: 10.1046/j.1525-1470.2002.00116.x.

Mail id- almosawiAJ@yahoo.com