Vici Syndrome

Alternative Names
VICIS
Immunodeficiency with Cleft Lip/Palate, Cataract, Hypopigmentation, and Absent Corpus Callosum
Dionisi-Vici-Sabetta-Gambarara Syndrome

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Other congenital malformations

Incidence per 100,000 Live Births
0-1

OMIM Number
242840

Mode of Inheritance
Autosomal recessive

Gene Map Locus
18q12.3-q21.1

Description
Vici syndrome is a rare, severe, multisystem disorder indicated by cardiomyopathy, corpus callosum agenesis, cataracts, oculocutaneous hypopigmentation, and immune dysfunction. While these are considered the characteristic features of the disease, patients may also suffer from developmental delay, progressive microcephaly, hypotonia, seizures, breathing difficulties, and a failure to thrive. Ophthalmological abnormalities also involve ocular albinism, retinal hypopigmentation, and nystagmus. Facial dysmorphia may include hypertelorism, low set ears, cleft palate, and micrognathia.

The syndrome does not have a racial or gender bias. It is a congenital disorder and symptoms usually present in the first year of life. Prognosis is poor and most patients succumb to the disease in infancy. Diagnosis is based on the presence of the characteristic clinical features. Genetic analysis of the EPG5 gene can help confirm the diagnosis. While there is currently no cure for Vici syndrome, patients require treatment for their individual symptoms. This may include antibiotics for infections, intravenous immunoglobulin replacement for immunodeficiency, and anti-convulsants for seizures.

Molecular Genetics
Vici syndrome follows an autosomal recessive pattern of inheritance. It is caused by mutations in the EPG5 gene that is involved in the highly conserved autophagy pathway. Both homozygous and compound heterozygous mutations in the EPG5 gene have been associated with Vici syndrome. Most of these are truncating mutations that result in reduced EPG5 protein.

Epidemiology in the Arab World

Egypt
See Saudi Arabia > [Byrne et al., 2016]

Oman
See Saudi Arabia > [Byrne et al., 2016]

Palestine
See Saudi Arabia > [Byrne et al., 2016]

Saudi Arabia
Byrne et al. (2016) identified 30 families with members that have Vici syndrome. Of these, two families were from Saudi Arabia, two were from UAE, two were Israeli-Arabs, one was Egyptian and one was Omani. Most subjects in the cohort presented in the neonatal stage, mainly with hypotonia and breathing difficulties. Symptoms included callosal agenesis, cataracts, hypopigmentation, cardiomyopathy, immune dysfunction, developmental delay, progressive microcephaly, and failure to thrive. The disease was found to be relentless in progression and neurodegenerative. Several patients had a severe seizure disorder. Patients also showed a variable presentation of neuroradiological features, such as pontine hypoplasia, delayed myelination, and thalamic signal intensity changes. Many patients

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died in infancy and the median survival age was analyzed to be 24 months. A total of 39 different EPG5 mutations (mostly truncating) were discovered in the cohort. survival outcomes were found to be significantly better in patients with the p.Gln336Arg mutation rather than truncating mutations. Survival was also better in patients with compound heterozygous mutations compared to homozygous mutations.

UAE

See Saudi Arabia > [Byrne et al., 2016]

References

Byrne S, Jansen L, U-King-Im JM, Siddiqui A, Lidov HG, Bodi I, Smith L, Mein R, Cullup T, Dionisi-Vici C, Al-Gazali L, Al-Owain M, Bruwer Z, Al Thihli K, El-Garhy R, Flanigan KM, Manickam K, Zmuda E, Banks W, Gershoni-Baruch R, Mandel H, Dagan E, Raas-Rothschild A, Barash H, Filloux F, Creel D, Harris M, Hamosh A, Kölker S, Ebrahimi-Fakhari D, Hoffmann GF, Manchester D, Boyer PJ, Manzur AY, Lourenco CM, Pilz DT, Kamath A, Prabhakar P, Rao VK, Rogers RC, Ryan MM, Brown NJ, McLean CA, Said E, Schara U, Stein A, Sewry C, Travan L, Wijburg FA, Zenker M, Mohammed S, Fanto M, Gautel M, Jungbluth H. EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain. 2016; 139(Pt 3):765-81. PMID: 26917586.

Related CTGA Records

Ectopic P-Granules Autophagy Protein 5, C. Elegans, Homolog of

External Links

http://disorders.eyes.arizona.edu/disorders/vici-syndrome
http://liftinglila.blogspot.ae/p/vici-syndrome.html
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=1493
https://rarediseases.info.nih.gov/diseases/448/vici-syndrome

Contributors

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