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Cancer Prone Disease Section

Mini Review

Hemihyperplasia isolated

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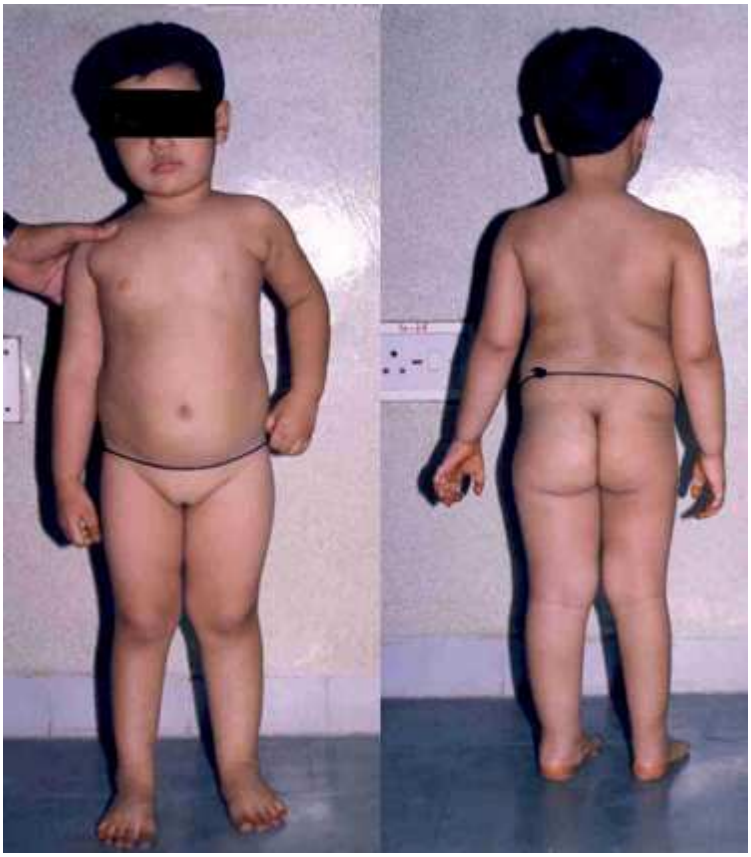
Identity

Other names: Isolated hemihypertrophy
Note: Excludes Proteus syndrome, Klippel Trenaunay Weber syndrome, neurofibromatosis, Beckwith Wiedemann syndrome, other syndromes associated with hemihypertrophy.
Inheritance: Sporadic.

Clinics

Usually hemihypertrophy occurs sporadically but familial cases are reported. Though molecular defects have not been identified in all cases, there is evidence that IH occurs due to epigenetic defects or paternal uniparental disomy of genes of 11p15 in somatic mosaic form.



A child with isolated hemihypertrophy involving left lower limb. Note Poland anomaly and hypoplastic nipple on left side - a rare associated feature.

Phenotype and clinics

Cases with hemihypertrophy not fulfilling criteria of complicated hemihypertrophies are grouped under isolated hemihypertrophy or hemihyperplasia. Though the title included the word 'hemi', only one limb may be involved. The condition is usually nonprogressive and the body disproportion does not change. Bone age may or may not be increased on the hypertrophied side.
Some cases of isolated hemihyperplasia have other features like naevi, capillary haemangiomas and hypertrichosis. Mental retardation may be present.There are no specific laboratory abnormalities. Viscera (kidney) on the hypertrophied side may be enlarged. Plexiform neurofibromas may look like hemihypertrophy.

Neoplastic risk

The risk of neoplasm is around 5%. Wilms tumour is the commonest; but other tumors like hepatoblastoma, adrenal cell tumour and leiomyosarcoma are also reported. Three monthly ultrasonographic follow up for Wilms tumour up to 5 years and then yearly up to the completion of growth is recommended.

Treatment

Usually limb discrepancy is mild and no treatment is required. Corrective shoes, orthopedic procedures may be needed to correct limb length discrepancy. Evaluation of cognitive function and appropriate training may be needed.

Prognosis

The condition is nonprogressive and the prognosis is good. Close follow up for early detection of Wilms tumour is needed.

Cytogenetics

Note: Mosaic paternal uniparental disomy for 11p15 (a region involved in Beckwith Wiedemann syndrome) was reported in an affected twin of a monozygotic twin pair discordant for hemihyperplasia. Paternal uniparental disomy of 11p15 in somatic mosaic form is reported in 16% of patients with isolated hemihyperplasia. Abnormal methylation of either LIT1 or H19 genes on chromosome 11p was seen in 8 out of 27 children with isolated hemihyperplasia; suggesting that isolated hemihyperplasia might belong to the phenotype spectrum of Beckwith Wiedemann syndrome. But there may be some differences in the types of epigenetic defects in both disorders. It has also been shown that the cases of isolated hemihyperplasia with uniparental disomy of 11p15 are at a higher risk of Wilms tumour as compared to the cases of hemihyperplasia without uniparental disomy.

References

Biesecker LG, Peters KF, Darling TN, Choyke P, Hill S, Schimke N, Cunningham M, Meltzer P, Cohen MM Jr. Clinical differentiation between Proteus syndrome and hemihyperplasia: description of a distinct form of hemihyperplasia. Am J Med Genet 1998;79(4):311-318.

Hoyme HE, Seaver LH, Jones KL, Procopio F, Crooks W, Feingold M. Isolated hemihyperplasia (hemihypertrophy): report of a prospective multicenter study of the incidence of neoplasia and review. Am J Med Genet 1998;79(4):274-278.

West PM, Love DR, Stapleton PM, Winship IM. Paternal uniparental disomy in monozygotic twins discordant for hemihypertrophy. J Med Genet 2003;40(3):223-226.

Niemitz EL, Feinberg AP, Brandenburg SA, Grundy PE, DeBaun MR. Children with idiopathic hemihypertrophy and Beckwith-Wiedemann syndrome have different constitutional epigenotypes associated with Wilms tumor. Am J Hum Genet 2005;77(5):887-891.

Martin RA, Grange DK, Zehnbauer B, Debaun MR. LIT1 and H19 methylation defects in isolated hemihyperplasia. Am J Med Genet A 2005;134(2):129-131.

Shuman C, Smith AC, Steele L, Ray PN, Clericuzio C, Zackai E, Parisi MA, Meadows AT, Kelly T, Tichauer D, Squire JA, Sadowski P, Weksberg R. Constitutional UPD for chromosome 11p15 in individuals with isolated hemihyperplasia is associated with high tumor risk and occurs following assisted reproductive technologies. Am J Med Genet A 2006;140(14):1497-1503.

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