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

This article should be referenced as such: