GATA-Binding Protein 3

Alternative Names
GATA3
Enhancer-Binding Protein GATA3

Record Category
Gene locus

WHO-ICD
N.B.: Classification not applicable to gene loci.

Incidence per 100,000 Live Births
N/A to gene loci

OMIM Number
131320

Mode of Inheritance
N/A

Gene Map Locus
10p15

Description
GATA3 is a member of the GATA family of dual zinc-finger transcription factors. This dual zinc finger transcription factor binds DNA with its C-terminal zinc finger (ZnF2) and stabilizes this binding with its N-terminal zinc finger (ZnF1). ZnF1 also interacts with other zinc finger proteins, notably Friend of GATA (FOG). The two zinc fingers of GATA3 are encoded by 2 separate exons highly conserved with those of GATA1.

Molecular Genetics
GATA3 is an essential transcription factor that was first identified as a regulator of immune cell function. In recent microarray analyses of human breast tumors, both normal breast luminal epithelium and estrogen receptor (ESR1)-positive tumors showed high expression of GATA3. Recent studies indicate that GATA3 may have a role in vertebrate embryonic development especially in the development of the inner ear. The hearing loss due to GATA3 haploinsufficiency has been shown to be peripheral in origin, but it is unclear to what extent potential aberrations in the outer hair cells (OHCs) contribute to this disorder.

Terminal deletions of chromosome 10p result in a DiGeorge (188400)-like phenotype that includes hypoparathyroidism, heart defects, immune deficiency, deafness, and renal malformations. One region that contributes to this complex phenotype is that for the hypoparathyroidism, sensorineural deafness, and renal insufficiency (HDR) syndrome.

Epidemiology in the Arab World
United Arab Emirates
Al-Shibli et al. (2011) described an Emirati boy who was diagnosed with HRD syndrome. Molecular analysis revealed a deletion mutation (c.35_36delGC) in the GATA3 gene in the patient. This was a novel mutation, and was not present in either of the healthy parents. The deletion was expected to cause a frameshift resulting in a prematurely truncated GAT3 protein or diminished GATA3 mRNA due to decay.

References
Al-Shibli A, Al Attrach I, Willems PJ. Novel DNA mutation in the GATA3 gene in an Emirati boy with HDR syndrome and hypomagnesemia. Pediatr Nephrol. 2011; 26(7):1167-70. PMID: 21399899

Related CTGA Records
Hypoparathyroidism, Sensorineural Deafness, and Renal Disease

External Links
http://genatlas.medecine.univ-paris5.fr/fiche.php?n=929
http://www.ncbi.nlm.nih.gov/gene/2625

Contributors
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