Non-syndromic Hearing impairment

Case of the Month Series 1101

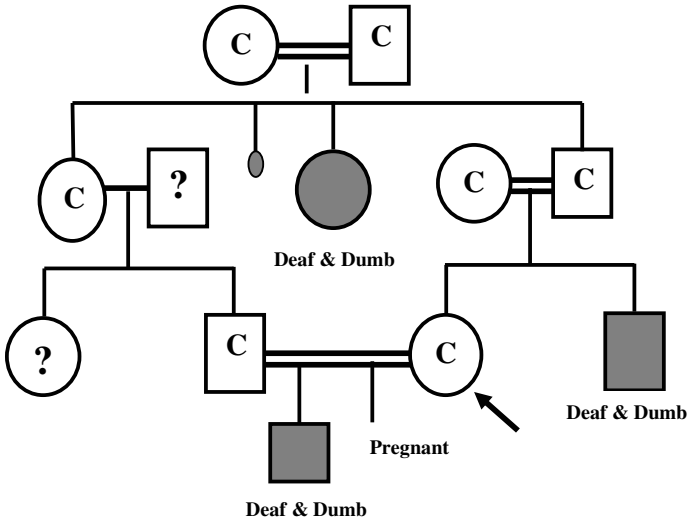
Genetics of Non Syndromic Hearing Impairment

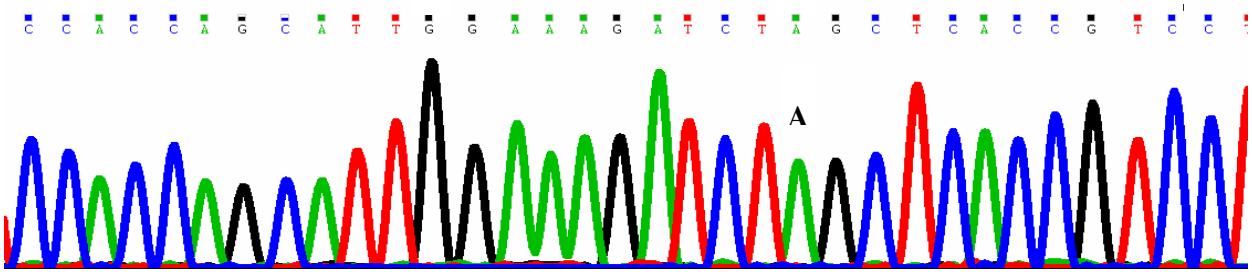
Non-syndromic (isolated) hearing loss occurs in 1/1000 children of which 50% have genetic causes & of those 50% are Autosomal Recessive type. The genes involved are GJB2 and GJB6 responsible in producing Connexin 26 and 30 proteins. Homozygous mutation (mutation on both DNA strands) in these genes results in hearing loss. It is common in consanguineous families. Parents are usually Heterozygotes (Carriers) of the gene mutations.

Case and counseling

A consanguineous couple with h/o a male child with hearing loss attended genetic counseling session in 2nd pregnancy at 10 weeks of gestation. Family history showed a clear Autosomal Recessive inheritance pattern of hearing loss in the family.

DNA testing of affected child by GJB2/GJB6 gene sequencing was recommended. The affected child showed G to A Homozygous mutation (also called W24X) on both DNA strands indicating that parents are carriers of the same mutation.

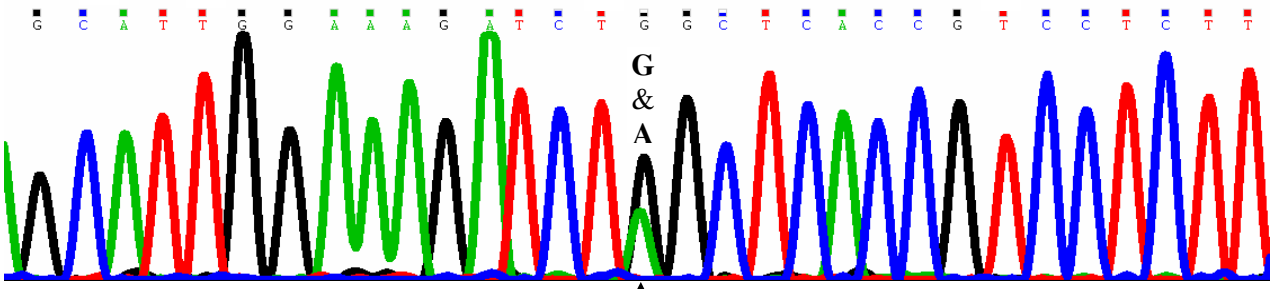




On both strands G was replaced by A

Prenatal Diagnosis

Amniocentesis was done at 16th week. DNA Sequencing of GJB2 gene of the fetus showed that the fetus was a heterozygote (carrier) for GJB2 G-A mutation (picture below) like parents. Therefore the fetus is unlikely to have hearing loss. The child is now 2yrs old, active and healthy with no hearing loss complaints.



G on one DNA strand and A on other

© Copyright, ATS GeneTech Pvt Ltd

GeneTech, First floor Amogh Plaza, Greenlands, Hyderabad, 500 016 **Tel**: 040-23400932, 23406178 **Fax**: 040-23407943
Customer Care: 98480-41127 (**AP**), 95909-41127 (**KA**), 9767641127 (**MH**) **Web**: http://www.genetech.co.in