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CapitalBio® Deafness-Related Gene Mutation Detection Kits

CapitalBio Technology
CapitalBio Genomics

Building C, 88 Kechuang 6th Street, Yizhuang Biomedical Park
Beijing Economic-Technological Development Area
Beijing 101111
China

Tel: +86-10-69002900 Ext. 8801
Fax: +86-10-69002988
Email: globalsales@capitalbiotech.com
Website : http://www.capitalbiotech.com

Background

A Worldwide Problem

According to WHO, over 5% of the world's population, that is, 360 million people, has disabling hearing loss. Among them, 328 million are adults and 32 million are children.

Hearing loss produces significant negative impact on individuals' life and the society. Individuals with hearing loss tend to have difficulty in communicating with others. Exclusion from communication can have a significant impact on everyday life, causing feelings of loneliness, isolation, and frustration. Spoken language development is often delayed in children with unaddressed hearing loss. Unaddressed hearing loss and ear diseases can have a significantly adverse effect on the academic performance of children. They often have increased rates of grade failure and greater need for education assistance. Adults with hearing loss tend to have a much higher unemployment rate. Among those who are employed, a higher percentage of people with hearing loss are in the lower grades of employment compared with the general workforce. As far as the society is concerned, WHO estimates that unaddressed hearing loss poses an annual global cost of 750 billion international dollars. This includes health sector costs (excluding the cost of hearing devices), costs of educational support, loss of productivity, and societal costs.

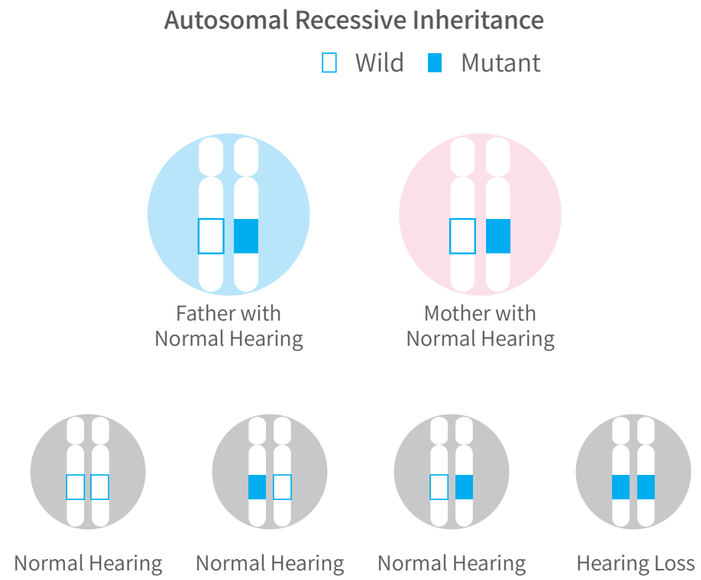
WHO points out that early detection and intervention are crucial to minimizing the impact of hearing loss on individuals and screening for ear diseases and hearing loss is an effective tool for early identification and management of hearing loss. Therefore, it urges member states to develop, implement and monitor screening programs for early identification of ear diseases and hearing loss in high-risk populations.

CapitalBio® Deafness-Related

Why Genetic Screening?

Autosomal Recessive Inheritance

The inheritance patterns of hearing loss include autosomal recessive, autosomal dominant, X-linked, and mitochondrial. The majority of genetic hearing loss is inherited in an autosomal recessive pattern and often presents in the absence of a positive family history for hearing loss. Statistics show that 95% of newborns with hearing loss identified by newborn hearing screening programs are born to hearing parents. That is, it is entirely possible for normal parents to give birth to children with hearing loss and whether a child's hearing is normal cannot be easily inferred from whether the parents' hearing is normal.



Late-Onset Hearing Loss

Not all hearing loss is present at birth. Hearing loss which is not present at birth cannot be detected by audiometric hearing screening conducted at birth. Genetic screening constitutes a useful tool for detecting mutations associated with late-onset hearing loss, thereby making early identification and management of late-onset hearing loss possible.

CapitalBio Fifteen Deafness-Related Gene Mutation Detection Kit

Description

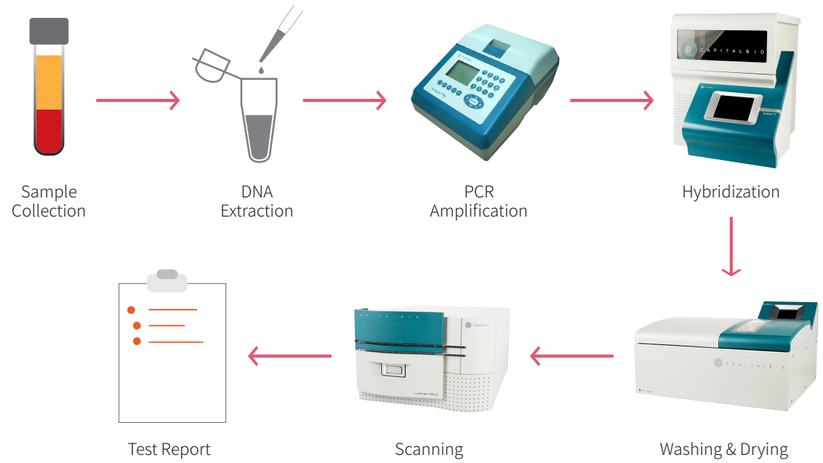
CapitalBio Fifteen Deafness-Related Gene Mutation Detection Kit is designed for a rapid and accurate detection of deafness-related gene mutations. It adopts the multiplex allele-specific PCR-based universal array (ASPUA) technology and covers 15 mutations in four genes.

Key Feature

High Throughput: As many as 15 deafness-related gene mutations are detected simultaneously.
Rapid Detection: It takes only 5 hours to finish the testing process.
High Sensitivity: Its detection limit is 3ng human genomic DNA/μL.

Gene	GJB2	GJB3	12SrRNA	SLC26A4
Mutation	35 del G 176_191 del 16 235 del C 299_300 del AT	538 C > T	1494 C > T 1555 A > G	1174 A > T 1226 G > A 1229 C > T 1975 G > C 2027 T > A 2168 A > G IVS7-2 A > G IVS15+5 G > A

Workflow



CapitalBio Genetic Deafness-Related Gene Mutation Detection Kit (Semiconductor Sequencing)

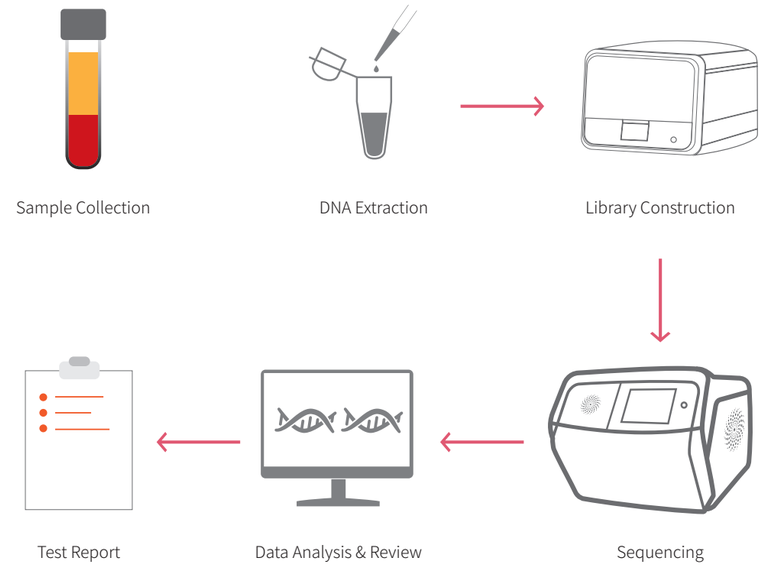
Description

Based on the NGS technology, the CapitalBio Genetic Deafness-Related Gene Detection Kit (Semiconductor Sequencing) is designed for a rapid, accurate, and comprehensive detection of genetic deafness-related gene mutations. It covers 100 mutations in 18 genes. It takes 5ml of peripheral blood or three dried blood spots only and the report can be finished in 2 business days.

Key Feature

High Throughput: As many as 100 deafness-related gene mutations are detected simultaneously.
Rapid Detection: The report can be finished in 2 business days.
High Accuracy: The accuracy rate ⩾ 99.9%.

Workflow



Mutations Covered by CapitalBio Genetic Deafness-Related Gene Detection Kit (Semiconductor Sequencing)

Gene	Mutation	Number of Mutations
GJB3	c.423delATT, c.421A>G, c.497A>G, c.538C>T, c.547G>A	5
DSPP	c.52G>T	1
GPR98	c.10088_10091delTAAG	1
DFNA5	IVS8+4A>G	1
SLC26A4	c.227C>T, c.230A>T, c.259G>T, c.269C>T, c.281C>T, c.334C>T, c.349delC, c.387delC, c.397T>A, c.404A>G, c.412G>T, c.439A>G, c.589G>A, c.679G>C, c.697G>C, c.707T>C, c.754T>C, c.812A>G, c.919-18T>G, IVS7-2A>G, c.920C>T, c.1079C>T, c.1105A>G, c.1115C>T, c.1160C>T, c.1174A>T, c.1181_1183delTCT, c.1226G>A, c.1229C>T, c.1318A>T, c.1336C>T, c.1343C>T, c.1540C>T, IVS13+9C>G, c.1555_1556delAA, c.1586T>G, c.1594A>C, IVS14+1G>A, IVS14-2A>G, IVS14-1G>A, c.1634T>C, c.1673A>T, IVS15+5G>A, c.1717G>T, c.1746delG, IVS16-6G>A, c.1919G>A, c.1975G>C, c.2000T>C, c.2027T>A, c.2054G>T, c.2082delA, c.2086C>T, c.2107C>G, c.2162C>T, c.2168A>G	56
TMC1	c.150delT, c.1334G>A	2
MYO7A	c.652G>A, c.731G>C	2
TECTA	c.4525T>G	1
DIABLO	c.377C>T	1
GJB2	c.512insAACG, c.427C>T, c.416G>A, c.299-300delAT, c.257C>G, c.253T>C, c.235delC, c.231G>A, c.218A>G, c.176-191del16, c.167delT, c.107T>C, c.99delT, c.94C>T, c.35delG	15
COCH	c.1535T>C, c.1625G>A	2
MYO15A	c.8767C>T	1
PRPS1	c.193G>A, c.259G>A, c.869T>C, c.916G>A	4
MT-RNR1	m.1494C>T, m.1555A>G	2
MT-TL1	m.3243A>G	1
MT-CO1	m.7444G>A	1
MT-TS1	m.7445A>G, m.7505T>C, m.7511T>C	3
MT-TH	m.12201T>C	1
Total		100

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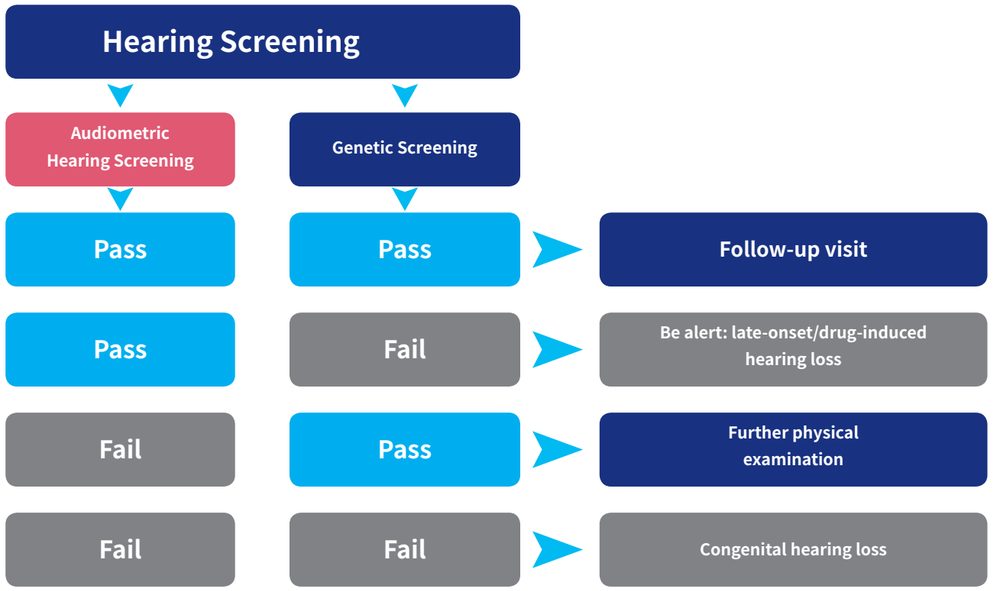
Screening Program

Newborn Screening



Prevention & Intervention