Mucopolysaccharidosis Gene List

سهامی خاص
شماره ثبت: ۴۱۰۴۵

Gene	Disease
IDUA	mucopolysaccharidosis type 1
IDS	mucopolysaccharidosis type 2
GNS	mucopolysaccharidosis type 3
HGSNAT	mucopolysaccharidosis type 3
NAGLU	mucopolysaccharidosis type 3
SGSH	mucopolysaccharidosis type 3
GALNS	mucopolysaccharidosis type 4
GLB1	mucopolysaccharidosis type 4
ARSB	mucopolysaccharidosis type 6
GUSB	mucopolysaccharidosis type 7
HYAL1	mucopolysaccharidosis type 11

نشانی: شیراز، خیابان انقلاب، خیابان شکیبا، روبه روی پارک انقلاب، ساختمان رز، طبقه اول، واحد۲ / کد پستی: ۷۱۶۳۷۶۶۵۱۵
تلفن: ۰۷۱-۳۸۲۶۳۰۶۹ / (۳۲۴۲) ۰۷۱-۲۳۰۵۸۸۶

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Importnat Notes:

1- Only known exons of these genes will be examined

2- Repeat expansion disorders will not be covered

3- Genomic regions beside exons of protein-coding genes, genes that are not listed here in this list, repeat expansions and mutations in the upstream and downstream regulatory regions will not be investigated.

4- Additional Comments:

- **Although next generation sequencing (NGS) is a method of choice for high throughput sequencing purposes, NGS has not been approved for clinical and diagnostic use; therefore, Sanger sequencing must be done to confirm the sequencing data, particularly on identified mutations.**
- **Genetic counseling is recommended to explain risks and potential**

5- pitfalls of the experiment.

- **It is of utmost importance for all clinicians involved in the care of families requesting molecular genetic diagnostic tests and the families themselves to be aware of the risk of errors in DNA analysis. Incorrect analysis may result from 1) incorrect data and clinical diagnosis 2) incomplete family studies and history 3) mix-up of DNA samples and mislabeling 4) rare molecular events5) new or spontaneous mutations 6) paternity problems, adaptation, IVF, egg donor, bone marrow transplantation, recent blood product transfusion 7) maternal DNA contamination of CVS or amniotic fluid samples 8) technical errors. The risk of errors from various reasons mentioned above and several others is about 0.5%, while the chance of technical errors of all types is estimated to be around 0.5%. The risk of errors due to DNA recombination in diagnosis is approximately 0.3%. We take no responsibility about patient identity and possible mis-labeling of the DAN samples. Any feedback from our colleagues in the clinical field would be most welcomed. Comments can be given in writing or by calling my number listed below or by e-mail to: Mohammad.ali.faghihi@gmail.com**

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