

Leber congenital amaurosis genetic testing

REQUIREMENTS FOR THE DNA SAMPLES

- The DNA quality needs to be ensured.
- 5 µg of genomic DNA is required for LCA chip analysis.
- Preferred concentration range of DNA is 100-250 ng/µl.
- DNA samples should be provided in pure sterile water.

TURNAROUND TIME

Express delivery – The results will be delivered in 3 – 5 working days after the arrival of samples.

Standard delivery – The results will be delivered approximately in 3 – 6 weeks after the arrival of samples.

OTHER TESTS PROVIDED BY ASPER

Asper Biotech	Asper Ophthalmics
Thalassemia	Stargardt disease
Cystic Fibrosis	Leber's congenital amaurosis
DNA repair	Usher syndrome
Hereditary Hearing Loss	Aut. Rec. Retinitis Pigmentosa
Ashkenazi Jewish	Aut. Dom. Retinitis pigmentosa
Wilson disease	Bardet Biedl syndrome
Breast and Ovarian canc.	Aut. Dom. Optic Atrophy
	Con. Stat. Night Blindness
	Corneal Dystrophy
	Vitelliform Macular Dystrophy

FOR FURTHER INFORMATION

- 1. Microarray-Based Mutation Detection and Phenotypic Characterization of Patients with Leber Congenital Amaurosis**
Yzer S, Leroy BP, Baere E, Ravel TJ, Zonneveld, VoesnekK, Kellner U, Ciriano JPM, Faber J-THN, Ro Suzanne Yzer, Bart P. Leroy, Elfride De Baere, Thomy J. de Ravel, Marijke N. Zonneveld, Krysta Voesenek, Ulrich Kellner, Jose P. Martinez Ciriano, Jan-Tjeerd H. N. de Faber, Klaus Rohrschneider, Ronald Roepman, Anneke I. den Hollander, Johannes R. Cruysberg, Franc,oise Meire, Ingele Casteels, Norika G. van Moll-Ramirez, Rando Allikmets, L. Ingeborgh van den Born, and Frans P. M. Cremers
Invest Ophthalmol Vis Sci. 2006 Mar;47(3):1167-76.
- 2. Microarray-based mutation analysis of the ABCA4 (ABCR) gene in autosomal recessive cone-rod dystrophy and retinitis pigmentosa.**
Klevering BJ, Yzer S, Rohrschneider K, Zonneveld M, Allikmets R, van den Born LI, Maugeri A, Hoyng CB, Cremers FPM.
European Journal of Human Genetics (2004) 12, 1024-1032.
- 3. Genotyping Microarray (Gene Chip) for the ABCR (ABCA4) Gene**
K. Jaakson, J. Zernant, M. Kulm, A. Hutchinson, N. Tonisson, D. Glavaci, M. Ravnik-Glavaci, M. Hawlina, M.R. Meltzer, R.C. Caruso, F. Testa, A. Maugeri, C.B. Hoyng, P. Gouras, F. Simonelli, R.A. Lewis, J.R. Lupski, F.P.M. Cremers, and R. Allikmets
Hum Mutat 2003, Vol. 22, pp. 395-403.
- 4. Genotyping microarray (disease chip) for leber congenital amaurosis: detection of modifier alleles.**
Zernant J, Kulm M, Dharmaraj S, den Hollander AI, Perrault I, Preising MN, Lorenz B, Kaplan J, Cremers FP, Maumenee I, Koenekoop RK, Allikmets R.
Invest Ophthalmol Vis Sci. 2005 Sep;46(9):3052-9.
- 5. Genotyping Microarray for the Detection of More Than 200 CFTR Mutations in Ethnically Diverse Populations.**
Schrijver I, Oitmaa E, Metspalu A, Gardner P.
J Mol Diagn. 2005 Aug;7(3):375-87

RECOMMENDATIONS FOR SHIPMENT OF THE SAMPLES

- For speedy and secure delivery, international courier services, for example DHL, UPS and FedEx, are recommended; alternatively, you can send samples by air mail.
- Since high quality DNA samples are stable, there is no need for shipment in dry or wet ice.
- Care should be taken to avoid drying out; please use either screw cap tubes or wrap the caps of each Eppendorf tube with parafilm.
- In order to avoid damage to the tubes during shipment, a tube storage box made of plastic or cardboard, and doubling it with a padded envelope, is recommended. Please avoid using round containers, such as 50 ml Corning tubes, for tube protection.
- Send samples to the following address:
Asper Biotech
Vaksali 17a
Tartu 50410
Estonia
Ph: +372 7 441 556
- Please fill in the DNA sample submission form (download the file from webpage) which improves and accelerates the handling of DNA samples and include it in the package.
- Notify us by email (info@asperophthalmics.com, or the respective project manager), including the number of samples, which test is to be performed and tracking data).
- Enclose in the package the list of samples, which test is to be performed and quality data, if available.
- Please make sure that the declared value for the package in the shipment documents does not exceed 10 EUR (USD).