



# Vitelliform Macular Dystrophy test

## REQUIREMENTS FOR THE DNA SAMPLES

- The DNA quality needs to be ensured.
- 1 µg of genomic DNA is required for VMD 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.

## 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 as a small parcel.
- 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 7307 295**
- 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](mailto: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).

## 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. **Evaluation of the Best disease gene in patients with age-related macular degeneration and other maculopathies**  
 Rando Allikmets, Johanna M. Seddon, Paul S. Bernstein, Amy Hutchinson, Andrew Atkinson, Sanjay Sharma, Bernard Gerrard, Wen Li, Michael L. Metzker, Claes Wadelius, C. Thomas Caskey, Michael Dean, Konstantin Petrukhin  
 Hum Genet (1999) 104:449-453
2. **Mutations in a novel gene, VMD2, encoding a protein of unknown properties cause juvenile-onset vitelliform macular dystrophy (Best's disease)**  
 Andreas Marquardt, Heidi Stöhr, Lori A. Passmore, Franziska Krämer, Andrea Rivera and Bernhard H. F. Weber  
 Human Molecular Genetics 1998, Vol. 7, No. 9, pp. 1517-1525.
3. **Allelic Variation in the VMD2 Gene in Best Disease and Age-Related Macular Degeneration**  
 Andrew J. Lotery, Francis L. Munier, Gerald A. Fishman, Richard G. Weleber, Samuel G. Jacobson, Louisa M. Affatigato, Brian E. Nichols, Daniel F. Schorderet, Val C. Sheffield and Edwin M. Stone  
 IOVS, May 2000, Vol. 41, No.6 (1291-1296)
4. **Bestrophin Gene Mutations in Patients with Best Vitelliform Macular Dystrophy**  
 Germaine M. Caldwell, Laura E. Kakuk, Irina B. Griesinger, Stacey A. Simpson, Norma J. Nowak, Kent W. Small, Irene H. Maumenee, Philip J. Rosenfeld, Paul A. Sieving, Thomas B. Shows and Radha Ayyagari  
 Genomics 58, 98101 (1999) Article ID geno. 1999.5808
5. **The mutation spectrum of the bestrophin protein functional implications**  
 Benjamin Bakall, Towa Marknell, Sofie Ingvast, Markus J. Koisti, Ola Sandgren, Wen Li, Arthur A. B. Bergen, Sten Andreasson, Tomas Rosenberg, Konstantin Petrukhin, Claes Wadelius  
 Hum Genet (1999) 104:383-389
6. **Identification of the gene responsible for Best macular dystrophy**  
 Konstantin Petrukhin, Markus J. Koisti, Benjamin Bakall, Wen Li, Guochun Xie, Towa Marknell, Ola Sandgren, Kristina Forsman, Gösta Holmgren, Sten Andreasson, Mihailo Vujic, Arthur A. B. Bergen, Valarie McGarty-Dugan, David Figueroa, Christopher P. Austin, Michael L. Metzker, C. Thomas Caskey, Claes Wadelius  
 Nature Genetics, Vol. 19, July 1998 (241-247)