



Testing for FSHD using epigenetics (Overview Pt 1)

Epigenetic testing for FSHD was developed by Dr. Takako Jones of the Peter and Takako Jones Lab in 2014 while at the University of Massachusetts Medical School. The technique was originally developed as a method to study the epigenetics of the FSHD region strictly for research purposes; however, it was soon realized that not only did the technique readily distinguish FSHD1, FSHD2 and healthy, it also addressed many of the problems associated with FSHD genetic testing. Thus, work began on transitioning this highly accurate yet simple and low-cost research method for use as a potential FSHD diagnostic. It is currently being used in the capacity of a research test to collect genetic and epigenetic information on FSHD worldwide and to provide information to individuals and families about their likely FSHD status.

Key paper describing the testing method

Jones et al. *Clinical Epigenetics* 2014, 6:23



METHODOLOGY

Open Access

Identifying diagnostic DNA methylation profiles for facioscapulohumeral muscular dystrophy in blood and saliva using bisulfite sequencing

Takako I Jones*, Chi Yan, Peter C Sapp, Diane McKenna-Yasek, Peter B Kang, Colin Quinn, Johnny S Salameh, Oliver D King, and Peter L Jones*

Key paper describing epigenetics in FSHD

Hypomethylation of D4Z4 in 4q-linked and non-4q-linked facioscapulohumeral muscular dystrophy

Nature Genetics (2003) 35:315-7

Petra G M van Overveld¹, Richard J F L Lemmers¹, Lodewijk A Sandkuijl^{1,5}, Leo Enthoven², Sara T Winokur³, Floor Bakels¹, George W Padberg⁴, Gert-Jan B van Ommen¹, Rune R Frants¹ & Silvere M van der Maarel¹

Advances in FSHD testing using epigenetics:

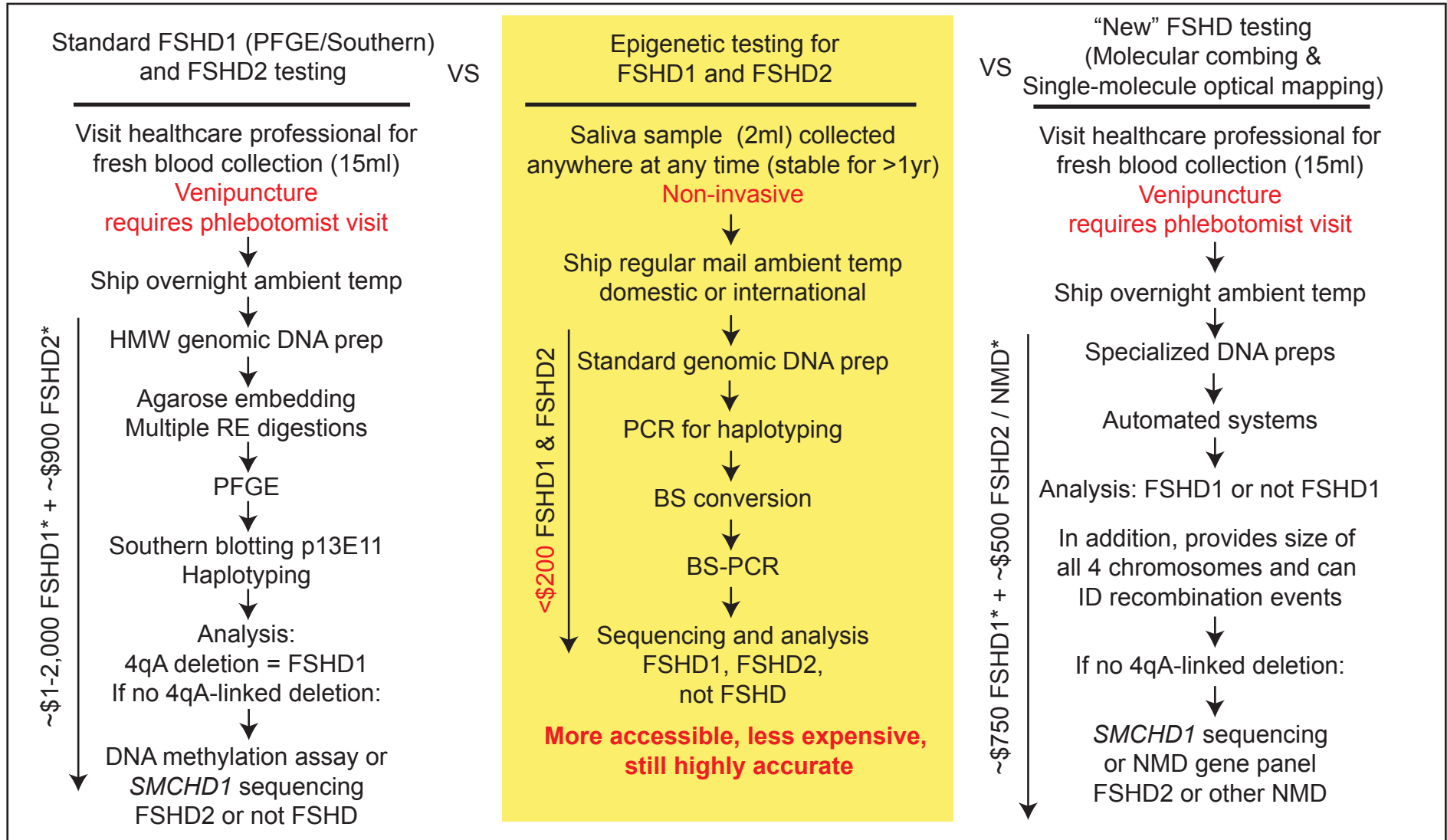
- ▶ Epigenetic FSHD testing can be performed on DNA from any human source (e.g., blood, muscle, saliva).
- ▶ Epigenetic FSHD testing can be performed on the same samples used for other DNA diagnostic tests (e.g., gene panels).
- ▶ Epigenetic FSHD testing identifies both FSHD1 and FSHD2.
- ▶ Epigenetic FSHD testing identifies D4Z4 contractions on nonpermissive alleles.
- ▶ Samples for epigenetic FSHD testing can be collected using a standard saliva collection kit sent through standard mail services to any location worldwide.
- ▶ Epigenetic FSHD testing greatly reduces the cost of FSHD testing.

Overall, epigenetic FSHD testing can make FSHD testing affordable and accessible to the worldwide FSHD community.



Testing for FSHD using epigenetics (Overview Pt 2)

Comparison of diagnostic workflows and cost



* These costs are estimates from information available at this time. Costs vary depending on company and technique and can change. Please see the individual testing providers for up-to-date pricing.



Testing for FSHD using epigenetics

(Overview Pt 3)

General flow for the epigenetic FSHD testing procedure.

Ref: T. Jones et al. (2014) *Clinical Epigenetics* 6:23

