



CLAIRE ALTMAN HEINE FOUNDATION, INC.

dedicated to identifying carriers of SMA

March 2009

CAHF Position Statement: Pan-Ethnic Population Based Carrier Screening and The SMA Patent

I. Introduction

The goal of the Claire Altman Heine Foundation (CAHF) is to achieve pan-ethnic population based carrier screening for Spinal Muscular Atrophy (SMA). SMA is building on the foundation formed in 2004 when standards of care were created for pan-ethnic population based carrier screening for Cystic Fibrosis. While CF and SMA are medically different, the pathway to instituting population based pan-ethnic carrier screening for SMA is remarkably similar to CF. In order to insure all couples of child bearing age are offered SMA carrier screening clinical access must be readily available. The enforcement and use of the intellectual property rights (i.e. the SMA patent) directly affects clinical access to SMA carrier screening.¹

II. Accessibility Issues

Carrier screening for CF is provided by over 65 different labs nationwide and over 100 academic labs nationwide and testing kits are readily available. While the CF gene is patented, broad, nonexclusive licensing practices have allowed for easy clinical access to testing, competition and innovation.

In contrast, carrier screening for SMA is only currently being performed by two labs (Athena and Genzyme). Licenses have not been granted for other laboratories or manufacturers of laboratory testing kits. Athena's aggressive enforcement and co-

¹ CAHF believes that genes and their mutations are naturally occurring substances that should not be patented. CAHF disagrees with the Patent and Trademark Office over the current enforcement of patents on genes that are important in the diagnosis, management and risk assessment of human disease.



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exclusive license with Genzyme of the SMA patent has resulted in an effective monopoly of genetic testing for SMA (both carrier screening and diagnostic). The patent for SMA is not so much the problem, but rather how the patent is being used and enforced resulting in a barrier to clinical access for SMA carrier screening.

As you know, aggressive enforcement of patent rights have many devastating effects:

- (1) Cause labs that might have offered SMA carrier screening to avoid offering these services thus limiting clinical access to testing.
- (2) Results in a lack of competition and incentive to improve testing services.
- (3) Eliminates competition for pricing.
- (4) Leaves no opportunity for independent confirmation of a test result (i.e. second opinion or full gene sequencing when needed).
- (5) Dictates and limits clinical access to both physician and patient.
- (6) Limits education of medical students and residents due to lack of exposure of patented material.
- (7) Limits the ability to use multiplex arrays, gene chips and other emerging technologies.
- (8) Hinders scientific and medical advancement of the field and diminishes accrual of knowledge about the molecular basis of the disease (by greatly reducing the number of laboratories with access to the gene).
- (9) Compromises quality assurance because the regular nationwide programs for proficiency testing (CAP/ACMG) are not interested in setting up these programs for a single customer.

III. Population based SMA carrier testing is becoming a “standard of care”

As carrier screening for SMA becomes a population based pan-ethnic carrier screen it is essential that the interests of medical practitioners and patients are taken into consideration during the licensing process. A sole or co-exclusive licensed provider of carrier screening for SMA is not in the best interest of the public health.

The needs of both physicians and patients in population based SMA carrier screening necessitates access to testing at several different facilities and the use of different testing modalities (like kits, gene chips, etc.). Most pre-conception and pre-natal testing is done in conjunction with other testing. The physician and patient want to eliminate the need for more paperwork in ordering testing, eliminate the discomfort and expense of extra blood collection, the “hassle” associated with insurance coverage issues and do not want



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to spend unnecessary time and energy on entering and negotiating license agreements for testing.

IV. Role of CAHF

CAHF expresses the concerns of the members of the general public, the carrier screening community, patients and clinical providers over the access problem with carrier screening for SMA. CAHF would like to play a role in mitigating the long-term access problems that result from exclusive licensing agreements. Most importantly, we want transparent, broad, nonexclusive licensing practices for SMA carrier testing so that every person has access to testing.

V. Licensing Solutions

The availability of models to secure licenses for SMA carrier screening, in reasonable practical and financial terms will encourage users (i.e. laboratories) to seek licenses and pay royalties. The licensing terms are often more important influences on accessibility than the existence of a patent.

CAHF would like to see licensing that includes:

- i. Transparency:
- ii. Clear Guidelines: License agreements should not provide the licensor with exclusive control. Athena should license SMA carrier testing on terms and conditions that seek to ensure the widest public access to, and variety of, products and services. Athena should license SMA carrier testing so as to be broadly accessible, at a fair and reasonable price that takes into consideration the different role(s) of academic and commercial labs. License agreements should not include up-front fees. In any case, licensing practices should not be used to restrict the choice of other products or services by patients and their health-care providers.
- iii. Non-Exclusive Licensing
- iv. Reasonable Royalties to avoid the effects of “royalty stacking” in pre-conception and pre-natal testing.



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VI. Conclusion

In order to achieve the clinical access needed for a population based pan-ethnic carrier screen for SMA, Athena and Genzyme must re-negotiate the terms of their co-exclusive agreement and take into consideration the needs medical practitioners and patients during the licensing process of the SMA patent.