

## MassARRAY(TM) System Enables Detection of Fetal Genetic Mutations in Maternal Plasma

Technology has the Potential to Replace Widely Used Risky and Expensive

### Diagnostic Procedures

SAN DIEGO, July 12 /PRNewswire-FirstCall/ -- SEQUENOM, Inc. (Nasdaq: SQNM) announced that through a collaboration with an international consortium led by The Chinese University of Hong Kong (CUHK) and Boston University (BU) a novel process to determine genetic mutations in circulating fetal nucleic acids in maternal plasma has been developed, as published in an article released in the online Early Edition of the Proceedings of the National Academy of Sciences. Investigators, led by Rossa W.K. Chiu, M.D., Ph.D., F.R.C.P.A. of CUHK and Chunming Ding, Ph.D. of BU, used MassARRAY technology to detect the inheritance of the four most common Southeast Asian beta-thalassemia mutations in at-risk pregnancies between 7-to-21 weeks of gestation and to analyze fetal haplotypes based on a genetic variation linked to the beta-globin locus HBB.

(Logo: <http://www.newscom.com/cgi-bin/prnh/20040415/SQNMLOGO>)

"Utilizing the superior sensitivity and specificity of the MassARRAY system, we have been able to isolate certain fetal point mutations and haplotypes in a non-invasive procedure with no risk to the fetus," said Charles R. Cantor, Ph.D., SEQUENOM's Chief Scientific Officer.

"SEQUENOM's ability to perform trace DNA analysis has enabled us to analyze circulating fetal DNA, a goal that on a large scale has been difficult in the past using other techniques due to their dependence on the fractional concentration of the circulating DNA and their lack of sensitivity. The MassARRAY system, however, is able to detect and analyze small amounts of DNA, even against the large background of maternal DNA. This new approach, in combination with our other offerings in various fields, shows the generality of mass spectrometry and its application to numerous areas of research and diagnosis."

"This development is very exciting and has addressed a long-awaited need in non-invasive prenatal diagnosis," said Y.M. Dennis Lo, D.M., D.Phil. of CUHK who discovered the presence of fetal DNA in maternal plasma in 1997 and who directs the international consortium. "In addition to beta-thalassemia, the platform can be readily applied to many other diseases of importance in prenatal diagnosis, e.g., cystic fibrosis. We also anticipate important applications in oncology where tumor-associated genetic mutations can be detected in the patient's blood plasma."

The method is based on SEQUENOM's MassARRAY technology and a new protocol called single allele base extension reaction (SABER), where primer extension is restricted to the fetal specific allele. The approach eliminates the significant risk of fetal loss associated with current invasive prenatal testing procedures, such as amniocentesis and chorionic villus sampling. In the US alone, about 200,000 invasive prenatal testing procedures are performed annually with associated costs of approximately \$200 million. The MassARRAY platform-supported approach is potentially applicable to other areas requiring genetic trace analysis such as early cancer detection.

### About SEQUENOM

SEQUENOM is a genetics company organized into two distinct business units: SEQUENOM Genetic Systems and SEQUENOM Pharmaceuticals. The Company has created high-performance DNA analysis technology and a platform that efficiently and precisely measures genetic variation. Both business units capitalize on this platform together with the Company's detailed knowledge of specific genetic variations in humans. SEQUENOM Genetic Systems is dedicated to the sales and support of the Company's platform, called the MassARRAY system, and to the continued expansion of DNA analysis applications for use with this system. SEQUENOM Pharmaceuticals has used MassARRAY technology and the Company's extensive collections of DNA samples from diseased and healthy individuals to identify disease-related genes that affect the health of significant portions of the population. The information from these studies is used for diagnostic and drug target identification followed by functional testing. The ultimate goals of the Pharmaceuticals business unit are diagnostic and therapeutic product development and commercialization.

SEQUENOM(R) and MassARRAY(TM) are trademarks of SEQUENOM, Inc.

Excent for the historical information contained herein the matters set forth in this press

release including statements regarding the potential of MassARRAY technology to replace widely used risky and expensive sampling procedures, the timing and application of the MassARRAY platform to other diseases of importance in prenatal diagnostics, in oncology and in early cancer detection, and the ultimate goals of the Pharmaceuticals business unit, are forward-looking statements within the meaning of the "safe harbor" provisions of the Private Securities Litigation Reform Act of 1995. These forward-looking statements are subject to risks and uncertainties that may cause actual results to differ materially, including the risks and uncertainties associated with SEQUENOM's technologies, and other risks detailed from time to time in SEQUENOM's SEC filings, including SEQUENOM's Registration Statement on Form S-3, most recently filed Quarterly Report on Form 10-Q, and Annual Report on Form 10-K for the year ended December 31, 2003. These forward-looking statements are based on current information that is likely to change and speak only as of the date hereof.

SOURCE SEQUENOM, Inc.

-0-

07/12/2004

/CONTACT: Linda Rawson, Corporate Communications Supervisor of SEQUENOM,

Inc., +1-858-202-9034, lrawson@sequenom.com/

/Photo: NewsCom: [http://www.newscom.com/cgi-](http://www.newscom.com/cgi-bin/prnh/20040415/SQNMLOGO)

AP Archive: <http://photoarchive.ap.org>

PRN Photo Desk, [photodesk@prnewswire.com/](mailto:photodesk@prnewswire.com)

(SQNM)

CO: SEQUENOM, Inc.; Chinese University of Hong Kong; Boston University

ST: California, Massachusetts, China

IN: MTC HEA BIO PUB

SU: PDT

AJ-CM

-- LAM007A --

1740 07/12/2004 06:30 EDT <http://www.prnewswire.com>

"Safe Harbor" Statement under the Private Securities Litigation Reform Act of 1995: Statements in this press release regarding Sequenom's business which are not historical facts are "forward-looking statements" that involve risks and uncertainties. For a discussion of such risks and uncertainties, which could cause actual results to differ from those contained in the forward-looking statements, see "Risk Factors" in the Company's Annual Report or Form 10-K for the most recently ended fiscal year.