

2 A Publication of the Coriell Institute for Medical Research, Volume 2, 2005

3 *In the three years since it started, the HapMap Project is already proving to be a powerful resource for researchers. Around the world, scientists believe that this valuable tool will help them to understand how genetics contributes to human health and diseases like cancer, heart disease, diabetes and depression, as well as aid them in finding the genes that affect people's responses to medicines.*

4 **First HapMap Milestone Reached**

With a big celebration and press conference in Salt Lake City, Utah, U.S. in October 2005, the International HapMap Consortium announced the completion of the basic HapMap. This resource, which includes a vast amount of data, shows the patterns of genetic variation in DNA samples from 270

people. The samples studied to develop this resource were from:

- ♦ Yoruba in Ibadan, Nigeria
- ♦ Japanese in Tokyo, Japan
- ♦ Han Chinese in Beijing, China
- ♦ CEPH (a Utah, U.S. community with northern and western European ancestry).

continued



“While many people around the world have contributed to the HapMap Project, we are especially grateful to those who donated their blood samples. Without their generosity, this project could not have been done.”

- Dr. Yusuke Nakamura, leader of the HapMap group at RIKEN and the University of Tokyo in Japan

6 About the Coriell Institute

The Coriell Institute for Medical Research in Camden, New Jersey is a not-for-profit basic research institute with an international reputation based on its accomplishments in genetics research and cell banking. Its cell repositories contain the world's largest cell culture collection for use in research, forming a central and irreplaceable resource for the worldwide scientific community.



7 How to Contact Us

We encourage your community, through your Community Advisory Group, to let us know what other types of information you wish to receive. At the Coriell Institute, Dr. Jeanne Beck oversees the Institute's involvement in the HapMap Project. Dr. Beck also coordinates outreach to participating communities and researchers. She can be reached at:

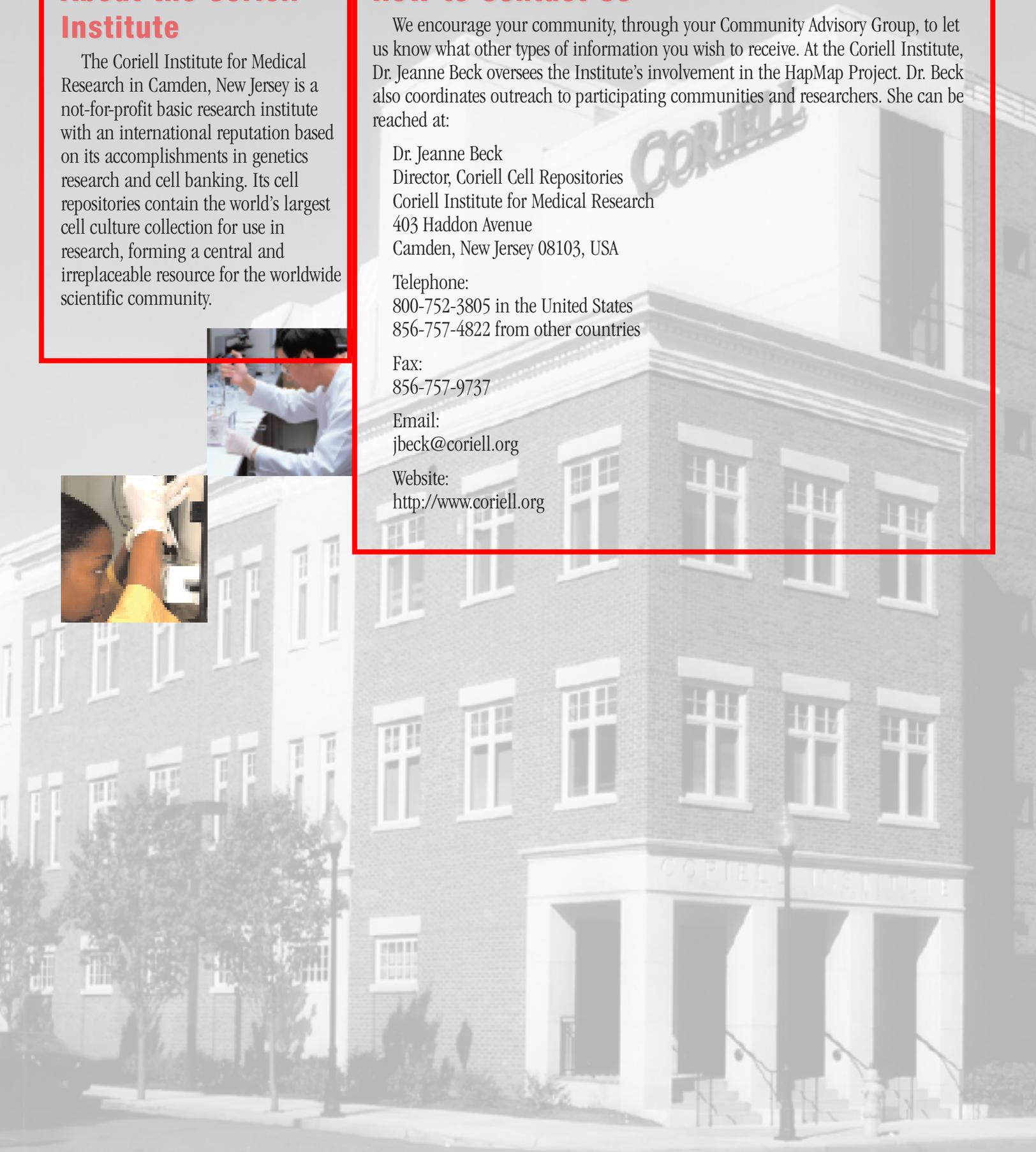
Dr. Jeanne Beck
Director, Coriell Cell Repositories
Coriell Institute for Medical Research
403 Haddon Avenue
Camden, New Jersey 08103, USA

Telephone:
800-752-3805 in the United States
856-757-4822 from other countries

Fax:
856-757-9737

Email:
jbeck@coriell.org

Website:
<http://www.coriell.org>



First Gene Discovered Using the HapMap

Even before the first phase of the HapMap project was finished, the HapMap was used to help find genes associated with people's risk for developing several common conditions. The first major success was the discovery of several genes related to age-related macular degeneration.

Macular degeneration affects 25-30 million people and is one of the leading causes of blindness in people around the world.

Of course, finding these genes will not provide an immediate cure for

macular degeneration. But it is an important first step. Based on this new knowledge, researchers may eventually be able to develop a test to see which people have the form of the gene (the "variant") that puts them at higher risk to develop this disease. When people are tested and have this variant, their doctors can advise them about extra precautions they should take to decrease their chances of blindness – like not smoking, staying out of bright sunlight as much as possible, eating more vegetables, and having more frequent eye

exams. Knowing about the genes related to macular degeneration will help doctors detect the disease earlier in people who already have it, and may someday lead to better treatment methods.

Other discoveries are on the horizon. For example, researchers are already using the HapMap data to help identify genes associated with type 2 diabetes, kidney disease, and the way different people respond to some drugs. Future issues of HapMap News will report on some of the new discoveries that will result from this and related research.

continued from page 1

"While many people around the world have contributed to the HapMap Project, we are especially grateful to those who donated their blood samples. Without their generosity, this project could not have been done," said Dr. Yusuke Nakamura, leader of the HapMap group at RIKEN and the University of Tokyo in Japan.

Reaching this milestone was just the first step in developing what will eventually become an even more useful resource. Starting soon, researchers will move into a new phase of the Project which will look at samples from seven additional populations:

- 10 ♦ Luhya from Webuye, Kenya,
- ♦ Maasai from Simba, Kenya
- ♦ People of African ancestry in the southwestern U.S.
- ♦ People from the Denver, Colorado (U.S.) metropolitan Han Chinese community

- 11 ♦ Tuscans in Tuscany, Italy
- ♦ People of Mexican origin in Los Angeles, California (U.S.)
- ♦ Gujarati (India) in Houston, Texas (U.S.).

12 Comparing the patterns of genetic variation in the samples from these groups will help researchers understand how well the basic HapMap, developed with samples from the first four groups, may work to find disease-related genes in people who live in or have ancestors from other parts of the world.

"As samples from more groups are studied, the HapMap will become an even more broadly useful and inclusive resource," said Dr. David Altshuler, leader of the HapMap group at The Broad Institute and Massachusetts General Hospital in Boston, MA (U.S.). While each community that donated blood samples for the Project is unique, all participating communities share a common feature: they have all made a major, lasting contribution to the future health of people everywhere.



14 *"As samples from more groups are studied, the HapMap will become an even more broadly useful and inclusive resource."*

- Dr. David Altshuler, leader of the HapMap group at The Broad Institute and Massachusetts General Hospital in Boston, MA (U.S.)

15 Special Feature

This issue highlights three participating communities

16 Han Chinese in Beijing, China

The Han Chinese population is the largest of 56 ethnic groups in China and the largest single ethnic group in the world. More than 90% of the Chinese population, and about 19% of the entire world's population, self-identify as Han. The name "Han" comes from the Han Dynasty, which ruled most parts of the geographical heartland of China from 202 BC to 220 AD.

The Han Chinese samples collected in Beijing were studied in Phase 1 and 2 of the HapMap Project. These samples came from members of the residential community at Beijing Normal University (BNU), a large, highly respected educational institution with 25 schools and departments, fourteen institutes, a night school, and continuing education programs. The residential community at BNU has almost 35,000 people, ranging from faculty and students to their families, university staff, and a wide range of professional and service workers. People from many different backgrounds were involved in discussions about the HapMap Project or donated samples for study, including people from 22 out of 31 Chinese provinces, autonomous regions, and municipalities.

The participants themselves understand that the HapMap Project will benefit the improvement of human health and medical research at large, and feel honored to have had the opportunity to contribute on behalf of China. Although there were some initial questions about the future uses of the samples, and some questions about where the samples will be stored and distributed, the CAG enjoys

16 cont. receiving information about the development of the Project itself and how the samples are being used by the larger scientific community.



17 According to members of the Community Advisory Group at Beijing Normal University (shown above) residents of this community were honored to contribute samples.

18 Denver, Colorado (U.S.) Metropolitan Area Han Chinese

Denver, Colorado, with a population of approximately 557,000, is home to 100,000 Asian people, of whom about 15,600, or 6.4%, self-identify as Chinese. The Denver, Colorado Metropolitan Area Chinese community is an especially heterogeneous community composed of people with many diverse emigration, social and cultural experiences. The people include those whose families immigrated to the area in the 1800's, exchange students studying in Denver in the late 1940's, individuals primarily from Taiwan and Hong Kong regions who came following the passage of the Immigration and Nationality Act

18 cont. Amendment of 1965 (which abolished national origin quotas and opened immigration), and people from Vietnam and the People's Republic of China who came in the mid and late 1970's, respectively.

The samples from the Denver, Colorado community will be studied in Phase 3 of the HapMap Project. Overall, community members and the CAG were eager to participate in the community engagement and sample collection activities. Many expressed hope that this genetic research will contribute to improving health and preventing diseases.

Members also reported that because they feel somewhat suspicious and cautious, they want genetic researchers to be respectful and truthful about their research as well as knowledgeable about the wide range of Chinese ethnic and cultural diversity, cultural values and practices without stereotyping individuals or their community. CAG members are looking forward to seeing how the samples collected from their community are being used in genetic research and look forward to their ongoing relationships with each other and the Coriell Institute.



20 The celebration of the Dragon Boat Festival is an important annual event for the Chinese community in Denver, Colorado.

21 CEPH (Utah, U.S. residents with northern and western European ancestry)

These samples, which were studied for Phase 1 and 2 of the HapMap Project, were collected in 1980 from people in Utah, U.S. whose ancestors mostly came from northern and western Europe. The name “CEPH” stands for the Centre d’Etude Polymorphisme Humain, the organization that originally collected the samples. The CEPH samples come from large, multigenerational families, and have been used in many other important genetic studies. Studying these samples for the HapMap will enable researchers to build on the extensive information about them that is already available.

Although some of the CEPH donors have died in the 25 years since the samples were collected, and some have moved to other places, most of the donors originally lived in an area

21 cont.

around Salt Lake City, Utah (population approximately 220,000). Salt Lake City is in a mountain valley and is famous for the Great Salt Lake, the largest salt-water lake in the western hemisphere. Salt Lake City is also the headquarters of the Church of the Latter Day Saints (also called the Mormon Church).

Although no formal CAG was established for the CEPH donors, most donors have maintained collaborative and trusting relationships with the researchers who collected the samples. The success of those long-term relationships, and the continuing scientific value of the CEPH samples as a reference collection, was an example that the HapMap Project sought to follow in collecting samples from other donor communities.



23

The CEPH collection includes multigenerational families from Utah, U.S. whose ancestors came mostly from northern and western Europe.

24 The next two issues of HapMap News will highlight other participating communities and continue to provide general information about the HapMap Project and its important discoveries.

25

An electronic version of this issue of HapMap News is available at <http://www.coriell.org/ccr/hapmap.html>. Additional information of interest can also be found on the website of the International HapMap Project, <http://www.hapmap.org>.

Continuing Community Involvement

27 To facilitate ongoing involvement by members of the HapMap donor communities, a Community Advisory Group (CAG) has been established at each site where samples for the Project were collected. The Coriell Institute, as custodian of the stored samples, welcomes suggestions from the CAGs about ways to improve the communication of information about the Project and the samples.

These suggestions will be useful even after the HapMap Project concludes, because the samples will be used both to develop the HapMap and for future genetic variation studies. "Although these future studies are unlikely to raise any new risks dif-

ferent from the risks raised by the HapMap Project itself, the Coriell Institute is committed to keeping the communities fully informed

about all such studies," said Dr. Jeanne Beck, the director of the Coriell Cell Repositories. To do this, the Coriell Institute distributes quarterly reports to each CAG, listing all the studies for which that community's samples have been distributed.



28 CAGs are also playing an important role in disseminating information to the broader local communities. Some CAGs use traditional methods of communication, such as informally discussing the Project with others in the community and distributing copies of *HapMap News*.

Some CAGs, in places where it is feasible, plan to create websites to post relevant information, such as CAG membership lists and minutes of CAG meetings. Publications about the HapMap and related studies, articles about ethical, legal, and cultural aspects of genetic research, and copies of *HapMap News* may also be posted.

29 *We hope, over time, to build an even stronger sense of the HapMap Project as a truly global enterprise among participating communities."*

- Dr. Charles Rotimi, leader of the HapMap group at Howard University in Washington, DC (U.S.)

30 The Coriell Institute will provide financial assistance to any CAG that wishes to maintain a website but lacks sufficient funds to do so. So far, one website has been established (<http://snp.ims.u-tokyo.ac.jp/CAG/JCAG/JCAG.html>) and the URLs for additional

30 cont. websites will be listed in future newsletters as they become available. They will also be available from both the Coriell Institute's website (<http://www.coriell.org>) and the International HapMap Project website (<http://www.hapmap.org>). It is hoped that this assistance will make it possible even for people in communities with traditionally little access to web technology to become more actively engaged in the research, learn more about donor communities in other parts of the world, and perhaps eventually establish direct communication with people in other donor communities. According to Dr. Charles Rotimi, leader

of the HapMap group at Howard University in Washington, DC (U.S.), who collected the samples from Nigeria and Kenya along with local collaborators, "We

hope, over time, to build an even stronger sense of the HapMap Project as a truly global enterprise among participating communities."

