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OBITUARY

Leena Peltonen-Palotie: the human side of genetics

Professor Leena Peltonen-Palotie, Genetic researcher, 2009. Photo by Jukka Rapo, Academy of Finland.

Virtually every contemporary scientist recognizes that achieving scientific excellence requires more than spending enormous amounts of time in the laboratory: there is a need for time to forge collaborations, a need for time lobbying agencies for research funding, a need for time celebrating a discovery, a need for time educating, motivating and acting as a role model for budding scientists and a need for time exposing the lay public and political bodies to the virtues of the practice of science. Few scientists are good at all of these activities and fewer still seem to actually enjoy even some fraction of them. It is most unfortunate then that the genetics community lost one of its brightest, most charismatic and most socially adept scientists in Leena Peltonen-Palotie this March to bone cancer. Leena's charm, good nature and tireless advocacy for good science were as noteworthy as her achievements as a scientist and educator, which were evidenced in more than 500 publications and in the training of more than 70 PhD students.

Leena began her career when the field of human molecular genetics was in its infancy and DNA markers had not yet been used to genetically map a single trait. By the time of her death, however, the field had assumed a position at the center of biomedical research, making molecular medicine a reality. Leena was a leader in every aspect of this remarkable transformation: she made unique and enduring scientific contributions, trained an extraordinary number of students and postdoctoral fellows and worked tirelessly for organizations dedicated to genetics. She believed passionately in the power of genetic science to advance medicine (as exemplified by an essay entitled *Genomics and Medicine* co-authored by Victor McKusick and published in *Science* in the issue presenting the initial draft of the human genome sequence in 2001 (1).

It is fitting that we remember Leena in *Human Molecular Genetics*, for which she served as a member of the editorial board for more than a decade. It is hard to think of a scientist whose work epitomizes so well both the mission of the journal and its evolution in the nearly 20 years since its founding in 1992. She published 50 articles in *HMG* (if this is not a record, then it must be close to the record) including at least one in each year of the journal's existence. Her first *HMG* papers reflect the struggles of the field—associated with the absence of genetic mapping tools that we now take for granted—in seeking to molecularly characterize rare Mendelian disorders. When positional cloning methods ultimately became available and reliable, Leena was ready to exploit them: during the 1990s, her group mapped and/or cloned more disease genes than the combined lifetime efforts of different researchers at most major institutes. These discoveries had a major impact in bridging basic and clinical investigation in medical genetics as well as diverse disciplines including immunology, cardiology, neurology and orthopedics.

In the wake of her discoveries concerning rare Mendelian disorders, Leena was among the first leaders in human molecular genetics to turn attention to common complex disorders. Beginning in the mid-1990s, she published numerous articles—including several in *HMG*—applying genetic mapping methods to disorders such as schizophrenia, bipolar disorder and multiple sclerosis. These methods were extensions of the methods she had employed so successfully in identifying genes that influence rare Mendelian disorders. By

the time of her latest *HMG* papers, published recently and posthumously, large-scale human gene mapping studies have identified several hundred replicated associations involving common human diseases. It thus goes without saying that Leena's efforts to identify disease-causing genes via linkage and association mapping strategies not only paralleled, but in many ways anticipated, contemporary large-scale genome-wide association study (GWAS) initiatives.

In this context, perhaps the most impressive of Leena's efforts derived from her ability to engage the public of her native Finland in scientific initiatives. She managed to capture the imagination of the country by emphasizing its unique genetic heritage, and we, like many foreign scientists, have been amazed at the obvious pride taken by Finns in their genetic distinctiveness. Leena was for many years the most widely known scientist in the country. Her ability to convey the importance of genetic discoveries in simple terms and with flair must at least partially explain why the Finnish population was so willing to participate in genetic studies. In communicating clearly to the public the benefits to be gained from utilizing this heritage to advance science, she created a legacy that is bound to endure. In fact, the common thread connecting her first published articles to her last published articles is the employment of Finland as a system for genetic investigation. Leena was not the first to describe the 'Finnish disease heritage'—the 40 or so Mendelian disorders that are more prevalent in Finns than in other populations—but she did more than anyone else to bring this phenomenon to the world's attention, as noted in several of her publications (2). The combination of complete sampling of Finnish cases and exploitation of linkage disequilibrium around disease genes that accounted for all Finnish cases enabled these many gene identification efforts by a single group at a time when, in most of the rest of the world, the available genomic tools would still have made this a difficult if not intractable task (3). The rapid identification of so many genes, with phenotypes that were so precisely documented, meant that Lena's group was able to devote a large proportion of its effort to functional characterizations—an obvious, necessary, but difficult next phase in any gene mapping study, no less so in the current era than when Leena began working in this area.

After gaining notoriety for her creativity, drive and ability to motivate the Finnish lay public and human genetics research community as a whole, Leena was able to embark on and guide a number of additional major international initiatives. For example, in the late 1990s, Leena accepted the challenge of founding and chairing a human genetics department at the University of California, Los Angeles, which remains the only such department among the 10 campuses of the University of California system. She returned to Finland in the early 2000s, playing a leading role in the development of large-scale European research programs which have proved the value of cross-national cooperation in advancing the genetic understanding of common diseases. Notable examples include the GenomEUTwin initiative which harnesses the power of European twin cohorts and the ENGAGE project which is integrating European GWAS meta-analysis efforts.

Leena was a superb teacher and wonderful mentor. In fact, as with the attention she gave to the Finnish lay public, for some time it seemed as though Leena was single-handedly managing the graduate and postgraduate training of an entire generation of Finnish biomedical researchers (and non-Finns who would engage her and her colleagues for training). Those of us fortunate enough to have been invited to Helsinki as opponents for one of Leena's PhD students can attest to the fact that these were memorable events: not just because of the theatre of Finnish PhD examination and not just because her students always seemed so smart, so confident and so well-prepared, but because of the obvious strength of the bond, scientific and personal, between Leena and each of her students. Leena's teaching initiatives were also very much social events, making them even more invigorating to students and faculty alike. Many can probably recall Leena's short courses on human genetics put together in the 1990s in which lecturers and students might be rewarded for a hard day's work with home-cooked dinners at Leena's house that included any number of dignitaries, entrepreneurs and scientists; pre-arranged passes to the prestigious Finnish Sauna Society; compact discs with a selection of Leena's favorite music; or any of a wide variety of well-intentioned, well-thought-out gifts. To most, such activities—especially in the context of a graduate-level educational series—would seem to go well beyond the call of duty, but to Leena they came naturally. In fact, there is a *facebook* group being formed coined 'supervised by Leena' to catalog and discuss the experiences students and trainees shared with Leena—a clear testament to the positive way in which Leena impacted her trainees and students.

Not surprisingly, Leena was very much a defender of women in science. Leena was incapable of discrimination and took everyone on merit, but also appreciated the additional obstacles (domestic and institutional) that work to impede women's academic progression. As a result, Leena had a reputation as one who did whatever she could to help her female students, postdocs and colleagues overcome these obstacles. In addition, anyone who knew Leena well appreciated her balancing of career and family. In doing so, she had the steadfast support of her husband, the geneticist Aarno Palotie, her partner in science and in life. She was immensely proud of and devoted to her adult children Laura and Kristian. Over the past several years, she and Laura established a tradition of rendezvousing to watch the Oscars together—and last year Leena flew to New York to uphold this tradition, exemplifying her determination to live her life fully as long as she was able.

Commemorative postage stamp. Reproduced courtesy of Itella Corporation/Philatelic Center.

Finally, Leena's infectious personality also made her something of a scientific celebrity. She embraced this role, probably in part because of her nature but importantly also because she knew that her standing and credentials in academia would provide motivation for young people who would come to know her through her television appearances and the popular magazine articles written about her. For example, in 2004, the Finnish television show, Suuret Suomalaiset, listed Leena as one of the 100 greatest Finns of all time, and on International Women's Day in 2010, she was honored by her home country in the form of a commemorative postage stamp.

Although it is all too trite to say that losing Leena is a clear detriment to the human genetics community, it is indeed worth emphasizing that Leena's intoxicating kindness, enthusiasm for learning and invaluable contributions to her chosen field and society in general should serve as an inspiration to individuals of all walks of life.

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