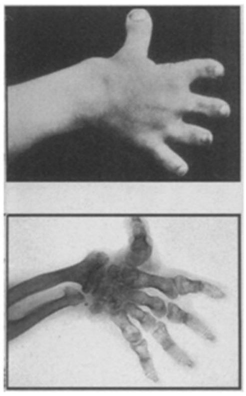
Biomedicine

New tool yields dwarfism gene . . .



Hästbacka *et al.*/Cell

This photograph and radiograph from a 1910 Russian thesis were the first to document the deformities of DTD.

It's been a good summer for little people. Scientists have now tracked down the genetic bases of two more skeletal abnormalities underlying short stature, information that can aid genetic testing and perhaps lead to the prevention of, or treatments for, dwarfism.

Most recently, a multidisciplinary U.S.-Finnish team used a new analytical technique called fine-structure linkage disequilibrium mapping to determine the cause of diastrophic dysplasia (DTD). DTD causes most of the dwarfism in Finland and is the third most common form in the United States. People who inherit two defective DTD genes develop twisted bones and arthritis, says Johanna Hästbacka of the Whitehead Institute for Biomedical Research in Cambridge, Mass.

Most of the 5 million Finns descended from a small group that 2,000 years ago settled what is now Finland. Thus, Hästbacka, Whitehead colleague Eric S. Lander, and Albert de la Chapelle of the University of Helsinki assumed that the defective DTD gene most likely arose from one person in that group.

The researchers first examined the genetic codes of affected members of 18 families with this disease and from that narrowed their search to an end of chromosome 5. They then analyzed the genetic material of 77 Finnish families having at least one member with a DTD gene, looking for ever smaller regions of that chromosome common to all these individuals. That common section represents the remains of the original ancestor's chromosome bearing this defective gene, Hästbacka says.

The DTD gene codes for a protein similar to ones already known to exist in rats and a fungus. Those proteins transport sulfate in and out of cells. Tissue taken from people with DTD does not take up enough sulfate, the group reports in the Sept. 23 Cell. Without sugar sulfate compounds, cells that make bone cannot lay down material that is both tough and resilient, she adds.

. . . while an old way pinpoints another

This summer, John J. Wasmuth of the University of California, Irvine, and his colleagues reported finding the gene that is defective in achondroplasia, the commonest genetic form of dwarfism in most parts of the world. That gene, called fgfr3, lies on one end of chromosome 4. Its protein product sits on the surfaces of cells and binds to specific growth factors, the group reported in the July 29 Cell. They found the gene quickly because they had first studied it as a possible candidate in Huntington's disease, Wasmuth says.

In achondroplasia, limbs are too short and the head is too large. A single amino acid change in the fgfr3 protein seems responsible, says Arnold Munnich of the Necker Institute at the Hospital for Sick Children in Paris. When he and his colleagues examined the DNA sequence of the fgfr3 gene in people with the growth disorder, they found that one nucleotide had been replaced by another. As a result, part of the protein that runs through a cell membrane contains an arginine amino acid where normally a glycine lies, Munnich and his colleagues report in the Sept. 15 Nature. They found this amino acid switch in six unrelated individuals with achondroplasia in their heritage and in 17 sporadic cases — in which parents spontaneously developed the genetic defect and passed it on to their children.

Computers

Technology more virtual than real

A medical student training to become a surgeon sits before a computer system equipped to enable her to see, hear, and feel a simulated, or virtual, heart. Without leaving her room, a teenager spends an evening shopping for a dress at her "synthetic environment" station, selecting fabrics and styles, then "trying on" the virtual outfits. An automotive engineer evaluates the control system of a new car by taking its virtual counterpart for a spin on a computer-mediated electronic highway.

Such are the dreams of researchers interested in virtual reality — systems in which a human operator can interact with a computer-generated world (SN: 1/4/92, p.8). But a National Research Council committee now cautions that a substantial gap exists between the technology available today and the technology needed to bring virtual environments closer to reality.

"Even the demonstrations of what are considered advanced [synthetic environment] research systems that can be seen at various universities, military installations, and industrial laboratories sometimes leave technically sophisticated observers who have no vested interest in the technology unimpressed," the panel asserts in its report, "Virtual Reality: Scientific and Technological Challenges." Nathaniel I. Durlach of the Massachusetts Institute of Technology chaired the committee, and Anne S. Mavor of the National Research Council served as study director.

A key issue concerns user comfort. Most applications of virtual reality to date have involved relatively short demonstrations, generally requiring bulky paraphernalia (especially headgear). If the comfort of this equipment can't be radically improved, "the practical usage of these systems will be limited to emergency situations or to very short time periods," the committee notes.

The panel also points out the need for more information about how people interact with virtual reality systems, especially when sound and touch are added to the visual milieu. For example, even in brief demonstrations of available systems, some people experience motion sickness or become disoriented. Very little is known about the psychological impact of this technology, particularly when the experience elicits powerful emotions from the participant.

Although much commercial development to date has focused on virtual reality systems for entertainment, the technology shows considerable potential in education and training, hazardous operations, medicine and health care, and design, manufacture, and marketing, the committee says. It recommends a wide range of research activities intended to help make virtual reality more practical.

"As with most other technologies, the effects of the advances in synthetic environments are likely to be mixed," the panel warns. "Some effects will be positive and others negative." By funding appropriate research, the federal government can influence the technology's future.

Lockheed

Wearing stereo goggles and a data glove, a researcher can "reach into" and interact in three dimensions with a computer-generated video display of acoustic data for antisubmarine warfare applications.