Muscular dystrophy defect located

Somewhere on a piece of one arm of the X chromosome lies an area responsible for Duchenne muscular dystrophy, the most common form of the muscle-wasting illness. Now, researchers report that they have pinpointed the area. The effort, led by Louis M. Kunkel of Harvard University, makes possible more accurate carrier identification and prenatal diagnosis.

Duchenne muscular dystrophy (DMD) is carried by mothers. Girls as a rule do not get the disease, because even if one of their two X chromosomes bears the deficiency, the other one can make up for it. But women with one deficient X chromosome are "carriers." Fifty percent of their sons inherit the faulty X and suffer the disease, which generally kills its victims before they reach their early 20s.

Ten researchers, from Harvard, Brandeis University in Waltham, Mass., Duke University in Durham, N.C., and the University of Pennsylvania in Philadelphia, report in the Aug. 29 NATURE the discovery of an X chromosome deletion at or very near the DMD gene, changing the scale of the genetic map from the range of millions to thousands of base pairs — the "rungs" on the DNA ladder.

The search began with the X chromosome of a boy who had muscular dystrophy and several other X-linked diseases. The scientists isolated the boy's X chromosome, which had multiple deletions, and the X chromosome from the cell line of a person who had four X chromosomes and did not have muscular dystrophy. They used enzymes to chop up the DNA, and they split the pieces from each set of chromosomes down the middle as though unzipping a zipper. The DNA halves from the cell line and from the boy were then mixed together. What didn't pair up — the piece of zipper half that had no mate — represented the DNA that was missing in the boy. When the extent of the missing DNA is fully determined, the researchers anticipate that the sequence will include the actual gene that, when disrupted, causes DMD.

The researchers used the unmatched pieces to test the DNA from 57 boys with DMD. On five of them, one of the pieces tested failed to match up, indicating that they, too, were missing chromosomal material in the same area.

The remaining 52 boys presumably had smaller genetic changes; inheritance patterns for their families can be determined by multiple genetic probes already devised by the researchers. These probes, which rely on knowing where on the chromosome the defect resides, employ "restriction fragment length polymorphisms," random variations in genes that can be used as genetic landmarks (SN: 8/31/85, p. 140). If the fetus, sister or daughter of a known carrier has the same polymorphisms in the DMD area that the carrier has passed to her DMD-affected son, there is a very high probability they also inherited the DMD gene.

The current work supplies the most exact map to date of the DMD gene. "We're at or very near where the gene is," says Allen D. Roses of Duke University. It improves upon recent work, including an international effort reported in the March 23 LANCET, that identified genetic markers. These markers, while better than the long-used but often-inaccurate muscle enzyme test, were still subject to error. They identified an area millions of base pairs away from the DMD gene, says P. Michael Conneally, a geneticist at Indiana University in Indianapolis, who wrote an accompanying commentary in NATURE. While they can be used to see if a person had inherited a defective chromosome, chance recombinations of genetic material reduce the accuracy. But the newly found location, Conneally told SCIENCE NEWS, "is right next door." And, he adds, "It's much closer to the gene, so it's a major breakthrough in finding the gene itself."

The probe is currently being used at several institutions in the United States and Europe to test relatives of boys with DMD, and both Conneally and Roses expect its use will become widespread for family screening. But it won't eliminate the disease. Up to a third of all cases are thought not to be inherited but instead to be caused by new mutations, and the diagnostic procedure isn't practical for mass screening. — *J. Silberner*

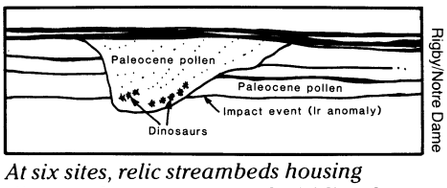
Could dinosaurs have survived asteroid?

The demise of the dinosaurs 65 million years ago has been unsuccessfully pegged to everything from poisoning and constipation to slipped disks and infertility. One of the most engaging ideas currently in vogue is that the dinosaurs were completely wiped out within a year or so after an asteroid slammed into the earth, spewing dust into the atmosphere that altered the climate and killed plants by blocking out the sun (SN: 6/2/79, p. 356). Now a paleontologist at the University of Notre Dame (Ind.) says he has evidence that challenges this theory too.

Keith Rigby and his colleagues recently discovered the bones of dinosaurs that they believe lived at least 40,000 to 200,000 years into the Paleocene epoch after the asteroid impact. The bones were found in streambed relics in the Hell Creek rock formation of east-central Montana.

The researchers do think there was an asteroid and they acknowledge its role in drastically reducing the number of dinosaurs; by their own reckoning the impact killed off at least 70 percent of these animals. But the asteroid did not deliver the final deathblow, they say. Their studies indicate that of the 13 dinosaur species living prior to the asteroid, 11 survived the impact. Moreover, the dinosaurs were already in decline before the asteroid hit. "The asteroid was the icing on a cake that had already been baked," says Rigby.

Rigby's group is not the first to suggest that dinosaurs lived beyond the crash. A few years ago, Jim Fassett, a stratigrapher at the U.S. Geological Survey in Reston, Va., reported what he believes are Paleocene dinosaur remains in New Mexico, which may have been left as much as 4 million years after the impact. Similar findings have been made in other places. And the impact theory has also been challenged on other grounds (SN: 8/31/85, p. 135). But none of these groups has been able to convince the scientific community of the validity of their claims. "This is the first time that there has been conclusive evidence that the dinosaurs did in fact survive the impact," says Rigby.



Rigby/Notre Dame

At six sites, relic streambeds housing dinosaur bones cut across the iridium layer left by the asteroid impact.

Rigby's findings have yet to be reviewed by other scientists because his group is just now submitting its complete results to SCIENCE. Fassett suspects that Rigby's most difficult problem will be to prove that the bones were not reworked—that is, not left during the Cretaceous period before the impact and then dislodged and reburied during the Paleocene. But Rigby says he has many pieces of evidence — such as dinosaur teeth with sharp edges that would have been worn down by extensive reworking — that support Paleocene origins.

Another uncertainty rests with the group's dating of bones, which was based on estimates of such inexact factors as sedimentary rates and changes in river flow patterns. Rigby notes that the dinosaurs could have lived even longer than his projections because the fossil record abruptly stops at a layer in which stream channels, the vehicle for collecting bones in high concentrations, disappear as the climate changes.

If Rigby's group is correct, then the question remains why the dinosaurs faced extinction while other species flourished. Rigby suggests that the dinosaurs' last days were spent along the banks of a Nile-like river with resources too limited to satisfy their greater needs. But the answer to "whodunit" must await the writing of the final chapter of this mystery.

—*S. Weisburd*