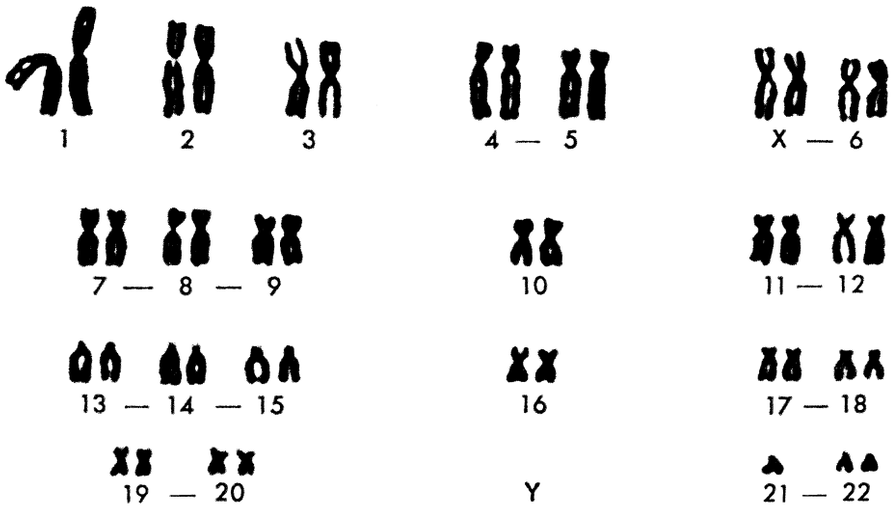

New England Journal of Medicine

Chromosomes of retarded girl: something missing in 21-22.

GENETIC ABERRATION

Survival despite autosome lack

A year ago, an Air Force technical sergeant and his wife, concerned over their three-and-a-half-year-old daughter's development, decided to seek help. The child could babble "ma-ma" and "da-da" but couldn't connect the sounds with her mother and father. She understood "no" but no other words.

The parents brought her to the Bethesda Naval Medical Center, where the National Institute of Child Health and Human Development maintains its Children's Diagnostic and Study Branch for the families of service men.

To the surprise of the researchers, as well as the parents, the cause of the girl's retarded behavior appears to be linked to a missing non-sex chromosome. The child is the first recorded case in medical literature to survive with this particular condition.

The specialists took the girl's complete history and performed tests including what is to them a routine chromosome analysis. She had only minor apparent physical abnormalities, and in most other clinics the tedious chromosome study would not have been undertaken.

Describing the child's appearance and characteristics in the Oct. 12 issue of THE NEW ENGLAND JOURNAL OF MEDICINE, the investigators say she had learned to play "pat-a-cake" at 18 months and would begin the game if the word was spoken—but only by imitation. Her usual play activity consists of sitting with her arms by her sides, rocking, or riding a hobby horse. She rocks her bed and bangs her head vigorously. Occasionally she will play with a toy car that has a siren, but not with more complicated toys.

The child's eyes are wide-set and downward-slanting, her mouth is small, her ears are low-set, and she has flared nostrils. She shows none of the characteristics of mongoloidism, caused by the presence of an extra chromosome.

Dr. Matti S. Al-Aish, who studies cellular genetics on the team that examined her, found that one of the child's autosomes—a non-sex chromosome—was missing. He believes it is probably chromosome 21, but he is not ready to commit himself further than to call it one of the Group 21-22.

The chromosome analysis was done by examining cultured blood, bone marrow and skin cells under the microscope until one cell was found in which the chromosomes were most clearly visible.

Chromosomes, which are present in all cells, contain the genes with the complete design for the living organism. One of the 23 pairs of chromosomes relate to sex; the other 22 pairs, the autosomes, determine other characteristics. Until this case was discovered it was believed that a missing autosome would cause death of the embryo in early stages of life.

Just what the missing autosome means in terms of missing characteristics is not yet known, and this unique case has aroused the interest of many geneticists. Dr. Al-Aish, with Drs. Felix de la Cruz, Lowell A. Goldsmith and Joseph Volpe made up the Institute team, assisted by Dr. J. C. Robinson, chief of the Pediatrics Outpatient Department of the Naval Medical Center.

Their report touches on possible causes of the child's condition, such as the parents' X-ray examinations without shielding and the mother's treatment for a cold while pregnant. The identity of the cold medicine is unknown, but during the last two months of the pregnancy she was given chlorothiazide for edema, or swelling, which often occurs in the ankles and feet at that time, though this is probably too late in the pregnancy to create the aberration.

"We believe," they say in conclusion, "this case illustrates the importance of cytogenetic studies, when they are feasible, in every retarded child." Studies now in progress could give further information about the missing chromosome's identity. ◈

24-HOUR DASH

Re-finding comets

Shortly after the beginning of the 18th century, astronomer Edmund Halley was comparing two dozen comet observations going back almost 400 years, when he noticed that the orbits of three comets—those of 1531, 1607 and 1682—were almost identical. The intervals between the sightings weren't quite the same, but the gravitational effects of Jupiter and Saturn could account for that. If they were indeed the same comet, Halley predicted, it ought to appear again in 1758. His guess was right on the nose: the comet, which now bears Halley's name, turned up, bright in the sky, as predicted.

If an astronomer is lucky, he may find a single new comet in his lifetime and have it named after him. A Japanese astronomer, Koichiro Tomita of the Tokyo Astrophysical Observatory, found a comet recently. In fact, he found so many—four—that he set a world's record for one evening's work.

Unfortunately, none of them will ever bear his name. They had all been found before. But Tomita is now apparently the champion comet re-finder of the world—at least for the 24-hour dash. (The lifetime record-holder is Dr. Elizabeth Roemer of the University of Arizona, whose 37 smother Tomita's 13.)

Relocating known periodic comets is a common procedure, valuable because the additional data from each new sighting makes it possible to predict the comet's next appearance more accurately. One of Tomita's rediscoveries, Comet Wolf, had been seen 10 times before since it was discovered by German astronomer Max Wolf in 1884. Tomita also made the fourth sighting of Comet Wirtanen, which was first seen in 1948; the third sighting of Comet Arend, known since 1951; and the eighth sighting of Comet Borelly, which was first found in 1905.

None of the four comets can be seen with the naked eye. The brightest of them is Wirtanen, which is of the 15th magnitude. The rest range down to magnitude 18, compared with Halley's comet, which is first magnitude and