Case Study – Human in Space
By Rachel Feltman

When astronaut Scott Kelly arrived in Houston on Thursday morning, he was about two inches taller than when he left for the International Space Station a year before, according to NASA representatives. That's pretty normal for an astronaut: Without the full strength of gravity pressing down on gel-filled discs between the vertebrae, they expand and lengthen the spine. It's a weird but temporary side effect of spaceflight.

But even if Kelly hadn't had his vitals checked immediately upon landing, he might have noticed the slight height change: One of the first Earthlings he saw was his identical twin, retired astronaut Mark Kelly — a man now notably, if only temporarily, shorter.

NASA scientists already knew that Kelly would walk a little taller when he emerged from the Soyuz capsule. But he'll have changed in other, less obvious ways, too — and that's the whole point of his recordbreaking mission. Kelly and Russian cosmonaut Mikhail Kornienko spent 342 days on the ISS to help scientists measure the effects of long-term spaceflight on the human body.

The impressive jaunt is a record for the United States, and for the International Space Station. But astronauts actually have stayed in space for months longer than that: Back in the pre-ISS days of Russia's Mir station, several cosmonauts broke this record.

But this is the first attempt to really study the effects of such a long stay in space. The hope is that data collected will highlight some of the most worrying physiological and psychological hurdles to spending months or years in space — allowing scientists to tackle those problems before NASA attempts a long-haul flight to Mars or the planets beyond. NASA hopes to put astronauts on the Red Planet by the 2030s.

Kelly's twin brother made him a perfect candidate for such a mission. The earthbound twin was used as a sort of "control" for experiments being run on his orbiting brother. Even identical twins aren't alike enough that NASA can pin every difference between them on the effects of spaceflight, but

finding changes that occurred in one man and not the other over the course of a year might give them some important clues.

“Obviously, this is a tiny sample size, so we’re not really looking at how Scott and Mark are different during the year, exactly,” Johns Hopkins Medical School's Andrew Feinberg told The Washington Post just before Kelly's launch last year. “It’s not statistically valid to say that differences between them must be due to the spaceflight.”

But, added Feinberg — whose project for the mission focused on epigenetics, or the way different environments affect the expression of our genes — “if something happens after Scott departs, increases during his trip, and then goes back to normal after he comes back to Earth — if we don’t see that kind of sequential change in his twin, well, it’s not proof of anything, but it certainly suggests something interesting is going on.”

The scientific experiments involved in the twin study are ongoing. All year, Kelly has been taking blood samples each time a shuttle is about to head back to Earth, allowing scientists to study fresh, unfrozen cells just hours after they’re drawn. Meanwhile, Mark has donated countless hours to providing samples of his own, as well as undergoing the same psychological and cognitive tests his brother completed in space. Now scientists can take all of that data and make something of it.

There are 10 official experiments running as part of the twin study, selected from a pool of research proposals submitted by institutions across the country: Six projects (including Feinberg’s) look at the way cellular behavior may change due to low gravity, high levels of radiation exposure, and other space-centric environmental factors. These studies will look at things like how aging, gene expression, and immune system function may have been affected.

Two studies will focus on physiological changes. One will try to determine whether plaque buildup in the arteries is accelerated during spaceflight, and another will look at how shifts in body fluid affect eyesight. Vision problems are a common complaint among astronauts.

Another study will use psychological tests to determine whether Scott Kelly has had changes in perception, reasoning, decision making and alertness that aren’t typical for someone spending a year on Earth.

And one last experiment will study how the microbes inside the twins’ guts have changed over the course of the year. Diet and environment can have a huge (and surprisingly swift) influence on the microbes that live in and around us, and there’s more and more evidence that these bug colonies have a profound effect on our health and wellness. Figuring out how bacterial colonies change in space could help keep the first Mars crew happy and healthy.

The hope is that scientists can come up with a plan for protecting the men and women who may eventually journey to Mars. It’s likely that the flight to our closest neighboring world will take about nine months each way. Once a crew touches down on the sandy surface, NASA will want them to stay awhile – after all, it would be a shame to spend 18 months in a tin can for a day or two on the surface of a new planet. So it seems likely that the first Mars mission will be a multiyear commitment.

“NASA is working on this science project that’s the greatest in the history of civilization,” Feinberg said last March. “They’re turning humankind from an Earth-dwelling species into a space-exploring species. One day, humankind will be a species that can settle on other planets. It might be a hundred years before we have humans living on Mars, but this is a whole new kind of science. It’s a multi-generational effort.”

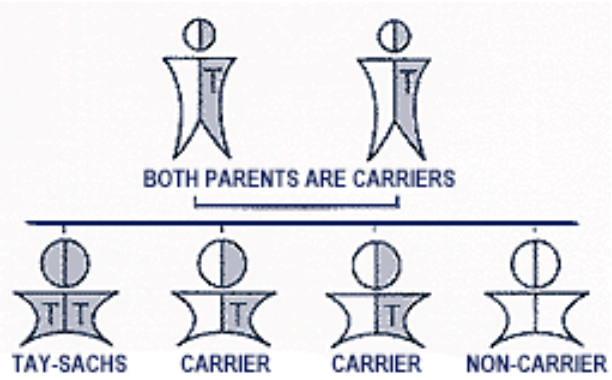
Case Study – Tay Sachs

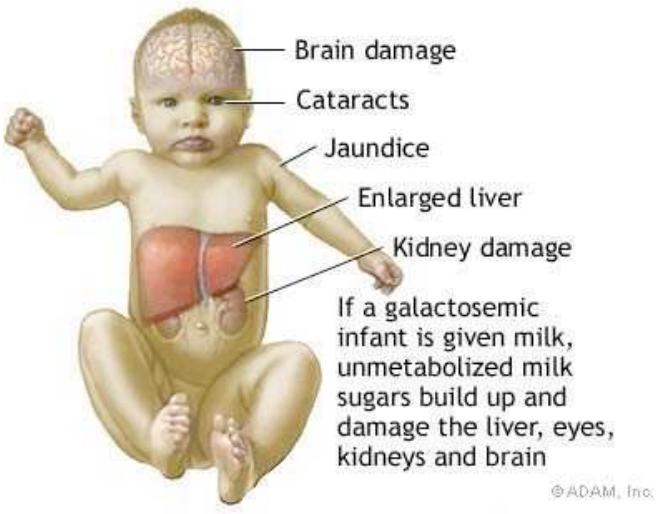
National Tay-Sachs & allied Diseases Association of Delaware Valley

A baby with Tay-Sachs disease appears healthy at birth, and seems to be developing normally for a few months. Symptoms generally appear by six months of age. While symptoms vary from one child to the next, there is always a slowing down of development. Gradually, Tay-Sachs children lose motor skills and mental functions. Over time, the child becomes blind, deaf, mentally retarded, paralyzed and non responsive to the environment. Tay-Sachs children usually die by age five.

Children with Tay-Sachs disease lack a vital enzyme, hexosaminidase A (Hex-A). Hex-A is needed for the body to break down a fatty waste substance found in brain cells. Without Hex-A, this substance accumulates abnormally and causes progressive damage until the nervous system can no longer sustain life.

Tay-Sachs is an inherited disease that only occurs when both parents carry a Tay-Sachs gene and each parent transmits the defective gene to their child. A child who inherits two Tay-Sachs genes (one from each parent) produces no functional Hex-A enzyme and is certain to develop Tay-Sachs disease.





A person with only one Tay-Sachs gene is perfectly healthy, but is a Tay-Sachs carrier. When both parents are carriers, there is a 1 in 4 (25%) chance, with every pregnancy, of having a child with Tay-Sachs disease.

When both parents are carriers, there is a 2 in 4 (50%) chance, with every pregnancy, of having a child who is a carrier.

When only one parent is a carrier, there is no chance the child will have Tay-Sachs disease. There is a 2 in 4 (50%) chance, with every pregnancy, of having a child who is a Tay-Sachs carrier.

Tay-Sachs carriers are found most frequently among families of eastern European Jewish descent (Ashkenazi Jews). In the United States today, approximately one in every 27 Jews is a Tay-Sachs carrier.

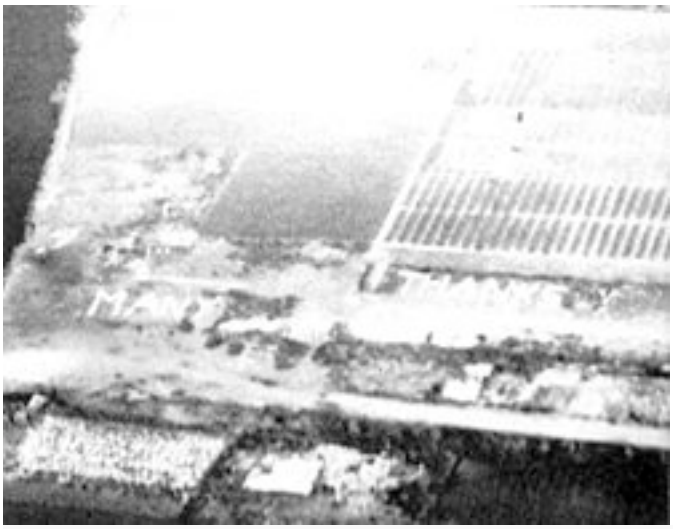
Among Jews of Sephardic origin and in the general, non-Jewish population, the carrier rate is about one in 250. There are certain exceptions. French-Canadian and the Cajun community of Louisiana have the same carrier rate as Ashkenazi Jews, one in 27. Also, individuals with ancestry from Ireland are at increased risk for the Tay-Sachs gene. Current research indicates that among Irish Americans, the carrier rate is about one in 50.

All couples planning to have children should carefully consider their ancestry to evaluate the risk of each partner. Any person who can trace his or her lineage to a high-risk population should be tested. In addition, close relatives of carriers (children, sisters, brothers, cousins, aunts, uncles) must be tested since they may also be carriers.

Case Study – Butterfly Wings

Light can influence gene expression, as in the case of butterfly wing development and growth. In 1917, biologist Thomas Hunt Morgan conducted studies in which he placed *Vanessa urtica* and *Vanessa io* caterpillars under red, green, or blue light, while other caterpillars were kept in the dark. When the caterpillars developed into butterflies, their wings showed dramatic differences. Exposure to red light resulted in intensely colored wings, while exposure to green light resulted in dusky wings. Blue light and darkness led to paler colored wings. In addition, the *V. urtica* butterflies reared under blue light and *V. io* butterflies reared in the dark were larger than the other butterflies.

Case Study – WWII and the Netherlands
BBC News



Operation Manna – "Many Thanks" written in tulips, Holland, May 1945.

People who were still developing in the womb at the time of severe World War II food shortages did worse than others of similar ages at mental tests almost 60 years later, researchers say.

Scientists, writing in the PNAS journal, said the 1944 Dutch "famine" may have accelerated brain ageing.

They studied nearly 300 adults who had been fetuses at the time.

UK experts said even severe morning sickness was unlikely to cause a similar level of malnutrition today.

The so-called Hongerwinter was a six-month period during which food deliveries to the people of the northern Netherlands were restricted by German occupying forces.

This produced a humanitarian disaster. By April 1945, it was estimated that 20,000 people had died as a result of malnutrition.

Many expectant mothers survived on between 400 and 800 calories a day.

However, the fact that this brief famine struck a previously reasonably well-nourished population allowed an almost unique opportunity for later scientists to study the effects of malnutrition on a group of children conceived around that time.

In this case, a group of almost 300 adults in their late 50s, all of whom had been exposed to the famine in the first or second trimester of their mother's pregnancy, were given mental tests, and the results compared to those of similarly aged people.

'Effective parasites'

This was the second time the group had been tested - tests in the 1970s had revealed no differences in performance.

However, in the second study, their results in a "selective attention test" were worse. Selective attention tests measure how well the brain can deal with competing distractions.

A classic example is a sequence of colour words, printed in different coloured ink, with the person asked to name the colour of each word.

There was nothing at birth to suggest a potential problem - as a group, their average birthweight was similar to babies whose mothers had not been exposed to famine.

Poorer performance in this type of test is generally linked to advancing age, and the scientists, from the University of Amsterdam and Calvin College in Michigan, US, suggested this might mean the brains of those in the study group had effectively started ageing faster as a result of malnutrition in the womb.

Dr Robert Fraser, an obstetrician based at Sheffield University, with a research interest in pregnancy nutrition, said that while the results were "interesting", they should not alarm modern mothers.

He said: "A baby is really a rather efficient parasite - a pregnant woman can be close to death from anaemia and the resulting baby born with a reasonably normal iron level in the blood."

He said that the severity of the Dutch famine meant that similar problems were highly unlikely for UK women.

"It was an awful, terrible time - people were scraping bin-lids with spoons, they were so desperate."

Dehydration

It is not unknown for modern women to be poorly nourished during the first and even the second trimester of pregnancy - the best-known cause being extreme morning sickness.

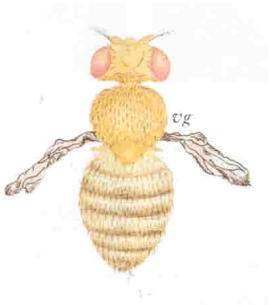
However, Fiona Ford, a dietician and spokesman for the British Dietetic Association, said: "The malnutrition would have to be pretty bad - with food intake at incredibly low levels, and there is evidence that the body is capable of adapting in these circumstances to protect the baby.

"If anything it is those women who are eating for two, or even three or four, who are more likely to cause a problem for their baby."

She said that, as a guide, women who lose 10% of their bodyweight or become dehydrated during pregnancy should consult their midwife or obstetrician.

Case Study – Drosophila

Vestigial winged fruit flies have small shrunken wings due to their homozygous recessive (vg/vg) genotype. However, flies that should have the vestigial phenotype will appear normal if they develop in an environment above 29° C. It appears that the high temperature turns-off the expression of the vg alleles.



Case Study – Snowshoe Hare

The snowshoe hare has genetic information for producing pigments that give its hair a brownish-grey color called agouti. However, during cold winter months the alleles for pigment production are turned-off and the hare appears white in color. In this case temperature regulates the expression of the coat color alleles.

Case Study – Hydrangea

Hydrangea flower coloration is subject to the pH of the soil in which the hydrangea bush is growing. In acidic soil, the flowers pink pigments are produced. In basic soil, blue pigmentation is produced in flowers.

Case Study – Hemophilia

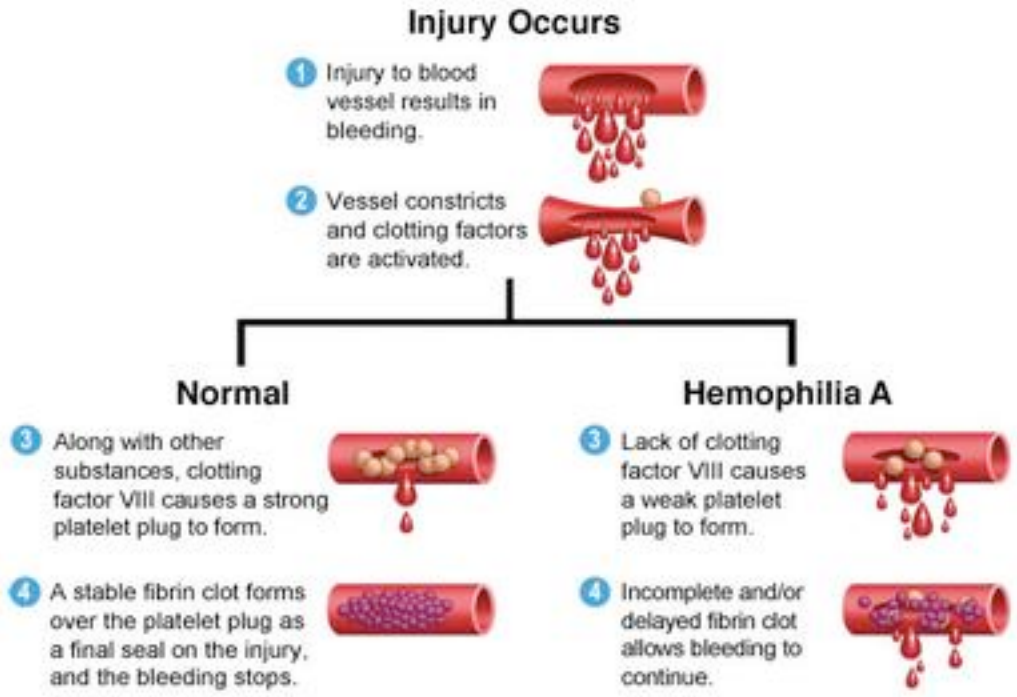
National Human Genome Research Institute

Hemophilia is a bleeding disorder that slows down the blood clotting process. People who have hemophilia often have longer bleeding after an injury or surgery. People who have severe hemophilia have spontaneous bleeding into the joints and muscles. Hemophilia occurs more commonly in males than in females.

The two most common types of hemophilia are hemophilia A (also known as classic hemophilia) and hemophilia B (also known as Christmas disease). People who have hemophilia A have low levels of a blood clotting factor called factor eight (FVIII). People who have hemophilia B have low levels of factor nine (FIX).

The two types of hemophilia are caused by permanent gene changes (mutations) in different genes. Mutations in the FVIII gene cause hemophilia A. Mutations in the FIX gene cause hemophilia B. Proteins made by these genes have an important role in the blood clotting process. Mutations in either gene keep clots from forming when there is an injury, causing too much bleeding that can be difficult to stop.

Hemophilia A is the most common type of this condition. One in 5,000 to 10,000 males worldwide have hemophilia A. Hemophilia B is less common, and it affects 1 in 20,000 to 34,500 males worldwide.



Case Study – Twins

Twins Separated at Birth Reveal Staggering Influence of Genetics
By Tanya Lewis, Staff Writer

Jim Lewis and Jim Springer were identical twins raised apart from the age of 4 weeks. When the twins were finally reunited at the age of 39 in 1979, they discovered they both suffered from tension headaches, were prone to nail biting, smoked Salem cigarettes, drove the same type of car and even vacationed at the same beach in Florida.

The culprit for the odd similarities? Genes.

Genes can help explain why someone is gay or straight, religious or not, brainy or not, and even whether they're likely to develop gum disease, one psychologist explains.

Such broad-ranging genetic effects first came to light in a landmark study — Minnesota Twin Family Study — conducted from 1979 to 1999, which followed identical and fraternal twins who were separated at an early age.

"We were surprised by certain behaviors that showed a genetic influence, such as religiosity [and] social attitudes," said Nancy Segal, an evolutionary psychologist at California State University, Fullerton, who was part of the study for nine years. "Those surprised us, because we thought those certainly must come from the family [environment]," Segal told Live Science. Segal described the groundbreaking research on Aug. 7 here at a meeting of the American Psychological Association.

Researchers at the University of Minnesota, led by Thomas Bouchard, launched the landmark study in 1979. Over the course of 20 years, they studied 137 pairs of twins — 81 pairs of identical twins (twins who developed from one egg that split in two), and 56 pairs of fraternal twins (twins who developed from two eggs fertilized by two different sperm).

The Jim twins were probably the most famous set of twins involved in the study, but other pairs were equally fascinating. One pair of female twins in the study were separated from each other at 5 months old, and weren't reunited until age 78, making them the world's longest separated pair in Guinness World Records.

The Minnesota study resulted in more than 170 individual studies focusing on different medical and psychological characteristics.

In one study, the researchers took photographs of the twins, and found that identical twins would stand the same way, while fraternal twins had different postures.

Another study of four pairs of twins found that genetics had a stronger influence on sexual orientation in male twins than in female twins. A recent study in Sweden of 4,000 pairs of twins has replicated these findings, Segal said.

A 1986 study that was part of the larger Minnesota study found that genetics plays a larger role on personality than previously thought. Environment affected personality when twins were raised apart, but not when they were raised together, the study suggested.

Reporter Daniel Goleman wrote in The New York Times at the time that genetic makeup was more influential on personality than child rearing — a finding he said would launch "fierce debate."

"We never said [family environment] didn't matter," Segal said at the APA meeting. "We just made the point that environment works in ways we hadn't expected."

Another study, commissioned by the editor of the journal Science, looked at genetics and IQ. The Minnesota researchers found that about 70 percent of IQ variation across the twin population was due to genetic differences among people, and 30 percent was due to environmental differences. The finding received both praise and criticism, but an updated study in 2009 containing new sets of twins found a similar correlation between genetics and IQ.

Moreover, a study in 1990 found that genetics account for 50 percent of the religiosity among the population — in other words, both identical twins raised apart were more likely to be religious or to be not religious, compared with unrelated individuals.

Other studies found a strong genetic influence on dental or gum health. That research helped to show that gum disease isn't just caused by bacteria, it also has a genetic component, Segal said.

Another study found that happiness and well-being had a 50 percent genetic influence.

In another study, researchers surveyed the separated twins about how close they felt to their newfound sibling. Among identical twins, 80 percent of those surveyed reported feeling closer and more familiar with their twin than they did to their best friends, suggesting a strong genetic component in the bond between identical twins.

The Minnesota study gave scientists a new understanding of the role of genes and environment on human development, Segal said. In the future, twin studies will aim to link specific genes to specific behaviors, as well as investigate epigenetics — what turns genes on or off, she said.

Segal, who wrote a book about the study called "Born Together — Reared Apart: The Landmark Minnesota Twins Study" (Harvard University Press, 2012), is now doing a prospective study of Chinese twins raised apart, often in different countries, by adoptive families.