

The Washington Post

HEALTH & SCIENCE E

TUESDAY, JULY 31, 2012

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**BEAR CAM**  
**In the wild, from your desk**Webcams let you watch Alaskan bears hunt and frolic. **E2****DOWN SYNDROME****Scientists look for drugs to aid mental abilities**  
One study shows small improvement in verbal memory. **E4****32**states and D.C. ban cellphone use by novice drivers. New federal incentives aim to reduce distracted driving. **Insuring Your Health, E4****“Instead of friends giving each other hugs, we're liking and poking each other via Facebook, and so we're all somewhat touch-deprived.”**A massage therapist, on the benefits of bodywork. **AnyBody, E5**

OWEN FREEMAN FOR THE WASHINGTON POST

An Air Force staff sergeant receives a misdiagnosis that affects her health and her life for decades

## What if she had known?

BY SANDRA G. BOODMAN  
Special to The Washington Post

To Jean McCathern, it seemed that the barrage of allergies — to pollen, mold, dust mites and cats — arose out of nowhere. In 1980 McCathern, then a 24-year-old Air Force staff sergeant assigned to the National Emergency Airborne Command Post at Offutt Air Force Base near Omaha, began gasping for breath as she carried equipment onto an aircraft.

**MEDICAL MYSTERIES**

Although McCathern had been in excellent physical condition, a doctor informed her a few weeks later that she had allergies and asthma, a diagnosis that remained unchanged for the next 24 years, even as her lung problems grew steadily worse.

McCathern's infirmities would cost her the career as a cryptographic repairman that she loved, lead her to part with her beloved cat and force her to give up one of her passions — attending hockey games — because the risk of illness from exposure to crowds was too great. It wasn't until 2004 that a simple blood test identified the malady that had plagued her for more than two decades — and it wasn't asthma or allergies.

“It was a pretty big shock,” said McCathern, soon to be 56, who lives near Harrisburg, Pa. Knowing earlier what was wrong, she added, “might have made a difference with what I did with my life.”

MYSTERY CONTINUED ON E5

## Genetic autopsies can help explain some sudden deaths

Medical examiners rarely order DNA tests, but they do reveal flaws and save lives

BY DARSIAK SANGHAVI  
Special to The Washington Post

By the time I first heard his name last summer, while working at my pediatric cardiology clinic at the University of Massachusetts Medical School, his face was all over the local television broadcasts. Josh Thibodeau, an active 12-year-old attending his first day of soccer camp in the town of Holden, had suddenly collapsed during a low-intensity passing drill. His younger brother Adam looked on in horror as emergency crews started chest compressions and tried to revive Josh with a defibrillator before rushing him to the

hospital.

Josh's parents, Deb and Ralph, arrived in the emergency room quickly, but they were too late. “The doctor came in to talk to me,” Deb told me recently. “It was very scripted, like on E.R.: ‘We have the best people.’ ‘We did all we could.’”

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Every year, researchers estimate, several thousand apparently healthy infants and children in the United States die suddenly and unexpectedly. Unlike with deaths in older people,

AUTOPSY CONTINUED ON E4

## ‘Mission of the decade’ nears Mars

BY MARC KAUFMAN



CURIOSITY ROVER ON MARS, AS DEPICTED BY NASA

washingtonpost.com

Watch the rover's landing sequence and an explanation of its instruments for exploring the Martian surface at [bit.ly/marsrover](http://bit.ly/marsrover).

The last time the United States landed a mission on Mars to look for extraterrestrial life or its building blocks, Gerald Ford was president and the nation had just finished celebrating its 1976 bicentennial.

Next week, the long-delayed second attempt will try to deposit a rover on the planet's surface.

The descent and landing in the early hours of Aug. 6 will be the most complex and hair-raising in planetary history. The destination is a deep crater with a three-mile-tall mountain that NASA could only dream about using as a landing site until very recently.

It's the most ambitious, the most costly (\$2.5 billion) and the most high-stakes mission ever to another planet. It was also described last week by the agency's top scientist, former astronaut John M. Grunsfeld, as “the most important NASA mission of the decade.”

“There is no doubt that this is a risky

MARS CONTINUED ON E6

## Federal money gives states extra incentives to shield young drivers from distractions

### Insuring Your Health

MICHELLE ANDREWS

Susan Valava's 15-year-old daughter, Kim, went to the movies one rainy night in 1995 with a 16-year-old friend who had just gotten his driver's license and two other kids. They weren't more than five minutes away from Valava's Wilmington, Del., home when the new driver, distracted by his friends' conversation, lost control of the car. Kim was killed instantly in the crash.

In most states today, such a driver would not be allowed to drive with a car full of friends so soon after getting his license and would have other driving restrictions, thanks to regulations implemented over the past decade. But safe-driving advocates say more needs to be done.

They hope that safety provisions included in the transportation bill signed into law by President Obama this month will encourage states to adopt or strengthen laws intended to protect teenage drivers — and everyone who shares the road with them.

It is an effort strongly supported by Susan Valava, who has worked in Delaware to stiffen regulations. "We knew [the driver] was a good kid. He'd been driving Kim to school every day the week before," she said. But the combination of "inexperience, a wet road, distraction and four kids in the car" led to tragedy.

Car crashes remain the No. 1 cause of death among teenagers, killing roughly 3,000 15- to 19-year-olds in 2009. Teens' lack of driving experience combined with their use of distracting devices such as cellphones make them the riskiest of drivers, four times more likely to crash as older drivers are, according to the Centers for Disease Control and Prevention and the U.S. Department of Transportation.

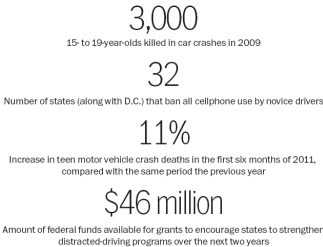
Every state and the District has its own graduated licensing program for teen drivers, according to the Insurance Institute for Highway Safety. The programs spell out the age at which a young person can get a learner's permit and generally require a certain number of hours of supervised training before a learner can take a driving test. After passing the test, teens enter an intermediate stage that allows them to drive without supervision but generally limits late-night driving and the number of passengers they can have in the car. These restrictions exist until the teen driver has reached a minimum age, often 17 or 18.

State programs vary considerably. In North Dakota, for example, a person can get a learner's permit at age 14, and there are no restrictions on the number of passengers an intermediate-stage driver can have in the car, according to the insurance institute. New Jersey residents, by contrast, must be at least 16 to get a learner's permit, and they can't drive unsupervised with more than one passenger in the car until they get their unrestricted license at age 18.

States have also moved to address distracted driving, a problem for both new and experienced drivers. Forty-four states and the District ban text messaging by novice drivers (39 states,



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including Maryland and Virginia, and the District ban texting by all drivers), and 32 states — including Virginia and, beginning Oct. 1, Maryland — and the District ban all cellphone use by novice drivers, according to the insurance institute.

After eight years of declining fatalities, teen motor vehicle crash deaths rose 11 percent to 211 in the first six months of 2011 compared with the same period the previous year, according to preliminary data from the Governors Highway Safety Association, which represents state highway safety offices.

The explanation might be as simple as an increase in the number of teens who are driving as economic conditions improve, says Barbara Harsha, the association's executive director. Others speculated that the increase might mean that current teen-driver safety programs may have achieved all they can.

And graduated driver licensing programs, even strong ones, may have their limitations.

A study published last year in the *Journal of the American Medical Association* examined the incidence of fatal crashes between 1986 and 2007 involving drivers between ages 16 and 19. It estimated that 1,346 fewer fatal crashes involving 16-year-old drivers occurred during that time because of graduated licensing programs than would have occurred without them. But the study also estimated that there were 1,086 more fatal crashes involving 18-year-old drivers over the same period, largely offsetting the improvements for younger drivers.

What's going on? One theory is that teenagers may be waiting until they turn 18 to get their license, bypassing the restrictions of the graduated licensing system altogether. They then have full license privileges, but they're as inexperienced as a 16-year-old.

"We know that new drivers have more crashes than more experienced ones," says John Uliczski, group vice president at the National Safety Council, an advocacy group.

Still, safety experts are encouraged that the new highway law provides \$46 million for incentive grants for states to implement or strengthen distracted-driving programs for all drivers over the next two years. It also includes \$27 million for states that adopt certain standards, including prohibiting cellphone use or communicating by device in non-emergency situations, for their graduated licensing programs.

This marks the first time the federal government has spelled out such standards. States can use the grant money for education, training and enforcement, among other things.

"We're offering a carrot" to states to strengthen their laws, says Jackie Gillan, president of Advocates for Highway and Auto Safety, an advocacy group. "This was a big step forward," she says.

This column is produced through a collaboration between The Post and Kaiser Health News, KHN, an editorially independent news service, is a program of the Kaiser Family Foundation, a nonpartisan health-care-policy organization that is not affiliated with Kaiser Permanente. E-mail:

## Stress may age brains of women

BY MICHAEL SLEZAK

Even though women live longer than men, their brains seem to age faster. The reason? Possibly a more stressful life.

As people age, some genes become more active while others become less so. In the brain, these changes can be observed through the transcriptome, a set of RNA molecules that indicate the activity of genes within a population of cells.

When Mehmet Somel, a computational biologist at the University of California at Berkeley, and his colleagues compared the transcriptome of 55 brains, they found that the pattern of gene activation and deactivation that occurs with aging appeared to progress faster in women.

"This was just the opposite of what we'd originally expected," says Somel.

He says that because women have longer lives, his group had expected to see slower or later aging-related brain changes. "But it fits every day observations on aging. Not all organs within an individual age at the same rate."

Somel's team compared the expression of more than 13,000 genes in four brain regions. In the superior frontal gyrus, which has been associated with self-awareness, the researchers found 697 genes that were expressed differently in men and women. Of those, 98 percent were skewed toward faster aging in women.

Some of these gene changes have been linked to general cognitive decline and degenerative disease.

The researchers noted that about half of the women showed accelerated age-related changes. They say that this suggests that the cause was environmental rather than simply biological. "A higher stress load could be driving the female brain towards faster aging-related decline," Somel says. His team found tentative support for that theory in a study of monkeys, where stress induced similar changes to their brain transcriptome.

Cyndi Shannon Weickert, a researcher at Neuroscience Research Australia, says the Somel group's findings are interesting, but the connection to stress is speculative. She notes that stress is only one possible cause of these effects. Inflammation, for example, might lead to similar changes.

health-science@washpost.com

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## Genetic autopsies can reveal threats to relatives

### AUTOPSY FROM E1

there often is no obvious cause, such as violence or overdose. Sometimes an autopsy detects an unsuspected condition such as brain bleeding, poisoning or infection. But otherwise, the fatality is labeled a case of sudden unexplained death. (In children up to age 1, the term is sudden infant death syndrome, or SIDS.)

It defies obvious solution. Observational studies from the 1980s showed that sudden deaths seemed more common in babies placed on their bellies. A national "back to sleep" campaign begun in 1994 led to an initial decline in infant deaths — perhaps in the fraction of fatalities caused by accidental suffocation — but for more than a decade now rates of unexplained sudden demise in infants and kids haven't budged. Without a clear diagnosis, families such as the Thibodeaus worry that another relative could drop dead at any minute.

Compounding the problem is that few of the thousands of medical examiners and coroners across the country are accredited, according to Andrew Baker, president of the National Association of Medical Examiners. Some of these examiners, their ranks can include elected officials without formal training — never look at the child's heart or brain in detail. Some perform X-rays and full toxicology tests in children who die suddenly; others check only for alcohol.

Nationwide, there is no consensus or regulatory guidance on how to autopsies on kids such as Josh, which makes it difficult for researchers to examine the phenomena of sudden unexplained death. "Our extended families lived in

fear," said Josh's mother, who has four other children and more than a dozen nieces and nephews.

### DNA provides proof

In 1998, Michael Ackerman, a researcher at the Mayo Clinic in Minnesota, heard about a 19-year-old athlete who had died after nearly drowning in only four feet of water while swimming at a fitness club in Iowa. At the time, Ackerman and several colleagues were studying a genetic disease called long QT syndrome.

Characterized by a hidden defect in certain cardiac cells, the syndrome had several bizarre variants.

One type results in cardiac arrest when patients hear sudden noises, like ringing from alarm clocks. Another caused a fatal rhythm problem when someone jumps into water. Suspecting that the Iowa victim had had that condition, Ackerman performed the world's first "genetic autopsy," using DNA extracted from her heart at autopsy, to show that she had long QT syndrome. He published the results in the *New England Journal of Medicine* and then determined that the woman's surviving sister also had the problem, which was treatable.

Thinking he was on to something, Ackerman got dozens of medical examiners in Minnesota to send him blood samples from sudden-death autopsies in young adults and children. He found that one-third had long QT syndrome or similar treatable genetic mutations affecting the heart. Roughly one-fifth of all SIDS cases in particular also showed it.

In another study, 30 percent of victims of unexplained drowning deaths positive for similar heart problems. (In

### What genetic tests can show

Genetic autopsies can detect several forms of the following causes of sudden death:

- long QT syndrome (eight types)
- Brugada syndrome
- catecholaminergic polymorphic ventricular tachycardia, or CPVT (two types)
- hypertrophic cardiomyopathy (more than a dozen types)

Reliable numbers are not available, but these tests are done in only a tiny fraction of cases.

Southeast Asia, researchers have found that another genetic cardiac condition, Brugada syndrome, causes many cases of sudden death.) A 2007 editorial in the *Journal of the American College of Cardiology* commented that genetic autopsy should "become part of the routine post-mortem study" in sudden unexplained deaths — not just to offer closure to families, but to save relatives from the same fate.

Though not directly involved in Josh Thibodeau's case, I was aware of Ackerman's research and asked the medical examiner handling his case to save blood for a genetic autopsy after I saw television reports about Josh's sudden death. Ultimately, Josh's body was taken to Boston for autopsy, and several tubes of blood were saved. After receiving consent from the Thibodeaus, our medical team helped the family send the tubes for DNA analysis.

### A matter of cost

A few weeks passed before the results were faxed to our office. The genetic autopsy showed a mutation causing excessive heart muscle growth and

rhythm disturbances — a disorder called hypertrophic cardiomyopathy. Microscopic testing of Josh's heart confirmed the diagnosis, and, reassuringly, additional genetic testing showed that none of his relatives carried the mutation. (Amazingly, however, testing showed that Josh's brother Adam has long QT syndrome, which was entirely unrelated to Josh's problem. Adam is now on a daily medication to prevent sudden death. He still plays competitive soccer.)

Because of the testing, Deb and her husband now know that they and their surviving children are safe. "Genetic autopsies aren't about the children who have died," Deb observes. "They're about those who were left behind."

Given how helpful such autopsies can be, why aren't more done? A large reason is the cost. In Josh's case, our team found a sympathetic claims reviewer at Blue Cross Blue Shield, who approved a genetic autopsy despite having no obligation to pay. But in most cases, once someone dies, health insurance does not pay for any further diagnostic testing. Labs charge \$2,500 to \$5,000 for the analysis, which the medical examiner's office must pay, roughly doubling or tripling the cost of an autopsy. As a result, none do the testing routinely in cases of sudden unexplained death and many never inform people of its availability.

I recently asked Henry Nields, the chief medical examiner for Massachusetts, how many genetic autopsies were performed among the dozens of children with unexplained sudden death in the preceding 12 months. To his knowledge, Josh Thibodeau was the only one.

### A determined stranger

Lisa Salberg, who runs the nonprofit Hypertrophic Cardiomyopathy Association, regularly scans news wires for stories of sudden unexplained death, and she calls medical examiners out of the blue to suggest genetic autopsies.

In 2009, a 14-year-old named Ian Artola was bare-knuckle sparring with a friend — one bystander called it a "little fight club" — at a New Jersey park when he collapsed and died.

On rare occasions, a blow to the chest from a karate kick or a baseball at precisely the wrong moment can disrupt the heart, causing sudden death. If that's what killed Ian, his friend would live with lifelong guilt. Hearing the story, Salberg called the county's medical examiner, and her group offered to fund a genetic autopsy on the teen.

"I know I sound like a stalker," Salberg tells me. "I'm calling them saying, 'I know you've got this body.' Ian's family consented to the test, and the results vindicated Salberg's hunch: The teen had the same problem as Josh Thibodeau. Further testing showed that his sister was fine but that his mother carried the gene. She is now monitored regularly."

No doctor or medical examiner or public health authority saved that mother's life. Instead, a determined stranger made a phone call. The problem, though, is that someone such as Salberg may not hear about the next case.

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**AUTOPSY CONTINUED ON E4**

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