

# ONE IN MILL- LION

Trisomy 18 is a rare genetic disorder that amounts to a death sentence. Meet 15-year-old Sarah Mah, living proof that nothing is certain

By **MALWINA GUDOWSKA** Photographs by **JARED SYCH**

**Sarah Mah** is not in the best of moods: her eyes roam around her classroom, every so often coming to rest on someone as she fixes them with an accusatory stare. Almost 16, she's just 45 inches tall (114 centimetres)—the size of a typical eight-year-old—but that intense look is at odds with her much younger appearance. With her long dark hair swept off her

petite face, wearing a bright purple top and black pants, she puckers her small lips and looks at me as if she can see what's hidden in my soul. All I can do is laugh nervously and apologize for tagging along during her school day. Her glare is off-putting to visitors, but to those who know her it is reassuring; it is evidence Sarah knows her own mind.

When Bev and Ron first saw the baby, the attending doctor began by saying things were just not right with Sarah. “We just thought, Oh, we can deal with that,” Bev says



**Sarah has** an extra 18th chromosome, a rare genetic disorder that causes a range of physical and developmental abnormalities, including brain and other organ malformations, and cardiac conditions. By all accounts, trisomy 18, as the condition is known, is a death sentence: it's almost unheard of for those born with the disorder to live longer than a year. But here she sits in her small wheelchair, trying to grab any hand in close proximity because she loves contact with people.

Until last year, Sarah often used a walker, but after surgery to correct a slipped kneecap she now moves around in a wheelchair because the pain in her knee is just too great. To communicate, Sarah uses noises—whining when she's irritated and laughing when something is right, it seems. Today, she adds the occasional laugh to her whines and disinterested turns of the head.

Even when she is in a bad mood, however, Sarah is living proof that there is an exception to every prognosis. Her mother, Bev Mah, is eager to make the most of Sarah's defiance of medical wisdom. "I want to expose Sarah to everything: to smell what a flower smells like, to feel a breeze in her face. I wanted to get her to feel as many sensations as she could, regardless of the time."

Bev and her husband Ron are kept busy around their Inglewood home attending to their daughter's needs. Sarah requires round-the-clock care and is in school full time.

**In Canada**, according to the Public Health Agency of Canada's *Congenital Anomalies in Canada—A Perinatal Health Report, 2002*, the fatality rate for severe anomalies such as trisomy 18 is virtually 100 per cent by the child's first birthday.

According to the Illinois-based The Support Organization for Trisomy 18, 13 and Related Disorders, 50 per cent of infants die within their first week of life and another 20 to 30 per cent die within the first month. The Canadian Organization for Rare Disorders states that trisomy 18 occurs in about one in every 5,000 to 7,000 live births and affects girls more often than boys. It is caused by spontaneous errors during the division of reproductive cells in one of the parents. Although no one really knows what causes these errors, the risk may increase with advanced parental age; Bev was 34 when she had Sarah.

Sarah's story begins in the customary way for children with trisomy 18, but the conclu-

sion is far from written. From the moment she was born at the Rockyview General Hospital, everyone expected Sarah to die at any moment. Indeed, just days after giving birth, Bev took Sarah to the funeral home to plan her funeral. "It's such a strange thing to grieve for your child when they are still alive, and that's how we lived for years—grieving," Bev says.

Sarah suffered from a number of problems commonly associated with her condition: frequent apneas (she would stop breathing because her brain could not control the function), slow postnatal growth and feeding difficulties. But each day she pushed on, leaving her parents, Bev, a communications consultant with the City of Calgary, and Ron, who runs his own catering business, to wonder if Sarah would be different.



The gang at Calgary Quest Children's Society: back row, left to right, Katarina Palanova, Sarah Palmer and Andrea Szabo; front row, left to right, Rachelle Beggs, Bev Mah with Sarah, and Becky Lampron.

**"It's such a strange thing to grieve for your child when they are still alive and that's how we lived for years—grieving"**

One day, Bev finally gave up waiting for her daughter to die, and set about giving her the best life had to offer. “When she was about seven, I said ‘I think she is going to live,’ and I really believed it. We just both started to change ... We weren’t scared anymore on a daily basis and we kind of allowed ourselves to be normal again,” she says.

Looking through the glass door of the Mah’s colourful home, the only sign that a disabled child lives there is the small wheelchair tucked against the wall. Bev, a small woman with reddish-brown hair, speaks slowly, pausing every moment over her cup of coffee. Her eyes are glossy, hints of tears appear, but she smiles as she recounts her daughter’s story. Ron is not home this morning; Bev describes him as a man who kept his distance from his daughter at first, paralyzed with the fear of getting too close emotionally, but who now can hardly bear being separated from her for any length of time. “She’s very like her dad—fathers and daughters,” Bev says. “She gazes at him like she’s looking at a movie star.”

In a later interview, Ron speaks about his daughter in the terms used by many proud fathers, often becoming speechless when trying to express his feelings towards her. “I just try to make her smile all the time whenever I see her,” he says. “She’s on the couch and I will kneel in front of her and she will grab you around the neck; it’s love without anything ... There is no hidden agenda—[it’s] just ‘I love you.’”

**Bev was unaware** during her pregnancy that Sarah had trisomy 18. But during a routine ultrasound at eight months, medical staff detected a cyst suggestive of a brain malformation (a Dandy-Walker malformation), but Bev was told not to worry about it. Although the doctor suspected something was wrong, she chose not to disclose her concerns since there was nothing anyone could do at that point. That is a decision Bev supports to this day.

When Bev and Ron first saw the baby, the attending doctor began by saying things were just not right with Sarah. “We just thought, Oh, we can deal with that,” Bev says. “Then he said, ‘Well, she won’t thrive,’ and I thought that meant she won’t run very well or keep up with the other kids ... He said, ‘No, she is not going to live,’ and it was such a blow. I can’t even remember the doctor’s face or anything that was around.” Sarah was Bev’s first child—Ron has two sons, Kevin and Chad, from a

previous relationship—and she had never changed a diaper let alone cared for a handicapped child.

The Mahs took Sarah home after being told that she would die within a month. “I lived in this environment where she was expected to pass quite quickly,” Bev says. “Every day it was a roller coaster of emotions because she would stop breathing and turn grey and blue, and sometimes I would try to give her CPR.” Ron, who returned to work, also faced the daily uncertainty. “Whether she was still there when I got home from work, that was always a question,” he says.

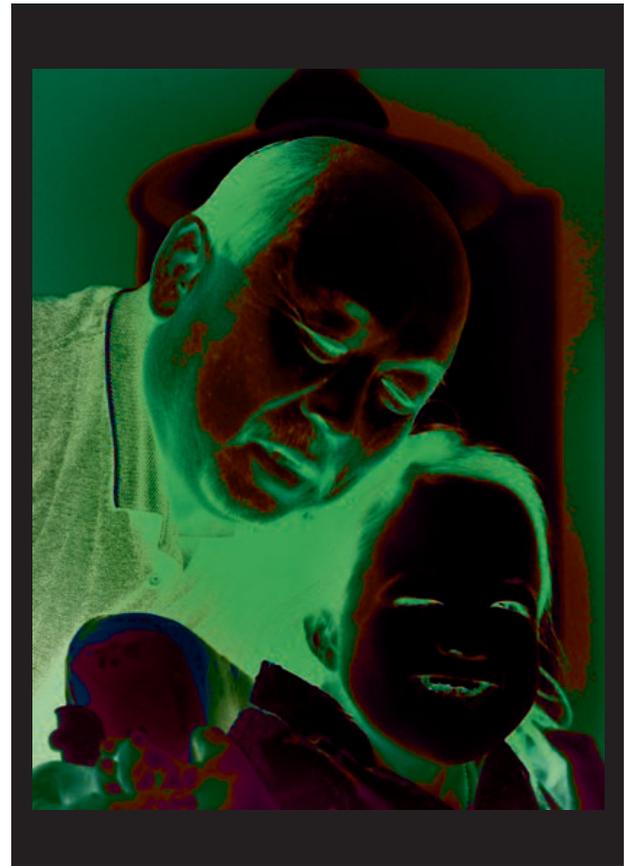
Each time Sarah stopped breathing, Bev would pat the child, not knowing if the jolt would help; each time Sarah began breathing again. But it wasn’t always that straightforward. “One day, [I was] washing the dishes and [the apnea] had happened and ... I lost it. Up until that time, I was just coping. ... I thought she was gone and I picked her up ... and I held her and I begged God—I didn’t believe in God but it was a good person to be angry with—‘Please take her; I can’t do this anymore,’” Bev says. “... I wanted her gone or [if] she’s gonna stay, stay. I couldn’t go with up and down anymore.” When Bev looked down again that day, pink spots were coming back into Sarah’s face. She was starting to breathe again.

Although Sarah has suffered complications such as seizures, poor vital signs and a major incident of intestinal bleeding, nothing has come close to the apneas of those first eight months. “Is it coincidence or is there someone who says you can only bear so much?” Bev asks philosophically.

**Sarah’s survival** qualifies as a medical mystery. Brian Kelly, a pediatrician in community practice, has only seen a handful of children born with trisomy 18, and yet two of his patients, including Sarah, have lived to be teenagers. Considering the statistics, two out of five is a high survival rate.

Kelly gave up many years ago attempting to explain how Sarah continues to live. “She had a ... very poor prognosis initially because her breathing was very erratic and she kept having multiple apneas,” he says. “She has overcome a lot of things, which, if [they] had not been promptly treated medically, could have gone the other way.”

Kelly says there is just no way to measure Sarah’s developmental abilities because there are no normal parameters. “It would be difficult to do cognitive testing. You tend to think



Like father. like daughter: Ron and Sarah share a laugh.

“When she was about seven, I said, ‘I think she is going to live,’ and I really believed it; and we just both started to change and we weren’t scared anymore on a daily basis. ... We kind of allowed ourselves to be normal again”

of her as a little baby because you carry her around and she is that size, but I know she looks around and is aware and thinks about what is going on and certainly has her own agenda sometimes," he says. "Just the way she carries on when she doesn't want to do something—she has her moods just like everyone else."

Everyone who knows Sarah says she has this mysterious ability to read people and

**"I lived in this environment where she was expected to pass quite quickly and every day it was a roller coaster of emotions because she would stop breathing and turn grey and blue, and sometimes I would try to give her CPR"**

situations. When Sarah's former caregiver visited the Mahs with her young child, the mother put the baby on the ottoman. Sarah was sitting in the room and suddenly began to make alarming noises, Bev followed her eyes and saw that the baby was slowly slipping off the ottoman. "We were so taken aback that she noticed this and she knew to tell us that [the baby] was in danger, so to speak, and we were just stunned," Bev recalls. "And this woman that was her caregiver at school was flabbergasted; she had no idea that Sarah had that ability."

Dr. Carey Johnson, a specialist in medical genetics sees two or three infants a year born with trisomy 18 in Calgary. (Because of the dire prognosis, many parents choose to abort, he says.) In his 20-year career he has seen only four long-term survivors, and he can give only a general, non-scientific reason for Sarah's longevity. "Some children are very resilient and robust . . . That allows them to go that extra mile and I give kudos to her parents," says Johnson. "She's lived this long, she obviously has some inner strength."

When Sarah and I meet for a third time, it is once again at her school, Calgary Quest Children's Society. Sarah is all smiles, she tries to grab my hand and laughs frequently. She is showing off, says Becky Lampron, who was Sarah's special needs teacher for two years and has been in the same classroom for six (every classroom has a number of teachers who each focus on two or three students). "She is like a normal teenage girl, she will let you know when she is not pleased with something," says Lampron.

Bev credits Quest with having greater ambitions for Sarah than she did. "It was the school that taught me how to interact with her," she says. "Frankly, when I started sending her to school . . . I thought, What are we doing this for? [The teachers] were the ones that said, 'We are going to teach her how to feed herself and she is going to say yes or no.' I thought, Yeah right, [but] they were so persistent and so creative in the things they

would come up with to teach her."

With each of Sarah's birthdays, the Mahs have become more confident that their daughter is here to stay. They began taking family trips with Sarah and her two stepbrothers, and they speak at conferences, offering support to other families affected by trisomy 18. "I feel like I am the person that pushes her around to these social events and I am just in the background . . . I watch her and people I don't even know come up to her," Bev says. "She just brings out wonderful things in people."

When asked what advice she would give other parents—with in-utero diagnosis, selective abortion is an option faced by many parents—Bev pauses for a long time. "I would say whatever you decide is right. This is fate and we can't change fate. Some children aren't going to be here long but you are lucky because you have felt their warmth and you have seen the colour of their hair and the colour of their eyes and some parents don't get to see that if the child is stillborn or they don't get to hear a voice."

Her comment reminds me of an incident that occurred on my second visit to the Mah's home. Sarah, Bev and I were sitting around the kitchen table—Sarah was eating a bowl of berry oatmeal and yogurt—when the phone rang. Bev answered and I knew she was talking about me: "She's still here, can I call you back?" After a pause, Bev laughed and said into the phone, "I think she already knows that," then hung up the phone and recounted the conversation for me: "He asked, 'Did you tell her Sarah is one in a million?'" It was Ron on the phone, adding all that he thought was necessary to his daughter's story. ■

Incomparable



*Alloy*  
HOMES

Design  
Construction  
Interiors  
Furnishing  
Real Estate

264 3667

www.alloyhomes.com