

LCC Session 19

CanMEDS Competency: Communicator

Dr. H Mijovic

What will happen in this session?

Physicians and other health professionals play an important role in how parents/caregivers conceptualize and come to terms with their child's medical diagnosis. Appreciation of parental/caregiver's thought process can be extremely helpful in building a long-term therapeutic relationship and determining goals of care.

During the first 20 minutes of this session you will be asked to read a short memoir "The Meaning of White" written by a mother whose infant daughter was diagnosed with oculocutaneous albinism.

As you read through the story, pay attention to the messages that Sadie's mother receives about her daughter's appearance and subsequently her daughter's diagnosis. What are these messages? Who are they coming from?

You will then be asked to share your thoughts about the story and work through the follow-up questions below.

Also enclosed in the reading package is a short article that looks at language choices in medical genetics, and the consequences for how individuals affected by the condition.

Suggested Time 60 minutes.

Readings:

E. Urquhart: "The Meaning of White" Walrus Magazine, April 2013. Pg 52-59.

J. Hodgkson et al.: "'SLANG"—Sensitive Language and the New Genetics—an Exploratory Study" Journal of Genetic Counseling, Vol. 14, No. 6, December 2005

Follow up questions for the group:

- 1. In your opinion, what is this story about? Was there anything in the story that you found particularly intriguing or surprising?**
- 2. How does Sadie's parents perspective on their daughter's condition change from the time of diagnosis until the end of the story when Sadie is a year old?**
- 3. What messages does Sadie's mother receive about her daughter's appearance and subsequently her daughter's diagnosis? Who are they coming from? What implications do they have for Sadie's parents?**
- 4. Based on this story and in your own experience, what are some of the messages should be conveyed to the family when their child is diagnosed with a genetic condition? Can you think of some effective strategies for communicating these messages?**
- 5. Sadie's mom is a PhD student in Folklore, her husband has a science background. She writes: " I am not certain that we are better off for knowing the molecular story rather than the folk tale, or whether there is room for both. Science can tell you how genetic anomalies and birth defects happen, but not why they happen to you rather than your neighbour. Medical facts can rarely offer the level of comfort that stories can."**
 - While Sadie's mom accepts her daughter's medical diagnosis, she also has alternative, non-medical explanations as to why her child is 'different'. Why do you think considering alternative explanations is important to her?**
 - Can you think of an experience from your practice where the family wondered about/ had an alternative explanation for their child's condition/illness (not necessarily genetic)? How did you come to know about their explanation? Did it in any way alter your communication with the family?**
 - Do you think it is important/helpful/relevant for the pediatrician to ask the family about their thoughts on why their child is 'different' or why their child became unwell?**

The Meaning of White

By Emily Urquhart • Illustration by Byron Eggenschwiler

Memoir

Science can explain why my daughter has a genetic disorder called albinism, but it is in stories that I find comfort

By [Emily Urquhart](#) • Illustration by [Byron Eggenschwiler](#)

March 20, 2013

From the [April 2013](#) magazine

THE WALRUS



The visitors come from all wards of the hospital. There is an audiologist, a social worker, a lactation consultant, a rotating cast of doctors, and an endless stream of nurses. We have a private room, but our newly formed family of three is rarely alone. This is not unusual in the maternity wing. What is curious, however, are the nurses who visit with no service to offer. They arrive at my side, somewhat apologetically, to catch a glimpse of our newborn daughter. “Some white,” they whistle and coo into her plastic bassinet, using the vernacular emphasis that has become so familiar during my five years in Newfoundland. They say it to me, and they repeat it to one another: “That hair is *somewhite*.”

Sadie Jane is born in the usual excruciating manner on Boxing Day 2010. Overdue, she is unwrinkled and chubby, with perfectly formed features and a shock of white hair on her head. Her mouth a tiny O and her arms flailing, she reaches constantly for my arms, my milk, my warmth. Her eyes flutter open occasionally, but mostly they’re shut. In a fleeting moment of wakefulness, the ward pediatrician probes her pupils with a tiny flashlight. Afterwards, she looks past me and my husband, Andrew, past my parents, fixing her gaze on the spruce-clad hills behind the hospital. “You have a very fair, very healthy baby girl,” she says. We never see the doctor again.

My child is the fairest of them all. The weight of my pride is unbearable, too big for our tiny room in the maternity ward. I stage a photo shoot on my bed, and Andrew takes the picture that will become Sadie’s birth announcement. I beam the image across the globe.

The next day, Andrew takes Sadie in his arms and goes for a walk down the hall. The nurses crowd around, making a fuss over her white hair and scolding him in the same breath. “No walking with babies in the hall! That hair! The liability!” He is heading back to our room when he overhears one of the nurses ask, “Is that baby an albino?”

They return trailed by a heavy-set nurse with dark hair and few teeth. “Is she an albino?” the nurse asks, lisping slightly, a note of alarm in her voice. “No,” I tell her firmly. The woman stares back at me, bug-eyed, bewildered. Then she lets herself into our bathroom, where she cleans the toilet, empties the trash bin, and wipes down the sink. She is wearing nurse’s scrubs, but it is clear now that she is a janitor.

When Andrew recounts this strange tale to his mother over the telephone, her heart sinks. She doesn’t know what to say, because both she and Andrew’s father, Don, asked the same question when they saw the first photographs of their granddaughter. Don, a family physician in Georgetown, Ontario, grows increasingly tense as the days pass. “Why didn’t the pediatrician say something?” he wonders. He is 99.9 percent certain, as convinced as he can be without examining her, that Sadie has a genetic condition called albinism. It is stable, and there is no treatment (you can’t substitute good genes for bad ones—at least not yet). He believes that the doctor opted to spare us, for now.

In a week’s time, Don will be on a plane to Newfoundland. His role as a grandfather is not to deliver grim medical news. He is the support staff, not ground control, and he feels certain that our family doctor will say something at the one-week checkup. After that, he can offer guidance.

Albinism, a genetic disorder, is both obvious and mysteriously complex. (As with the pejorative “retard,” those in the know don’t use the word “albino” anymore.) People with oculocutaneous albinism have little to no pigment in their skin, hair, and eyes. They have relatively little protection against the sun; burns are quick and dangerous and may cause skin cancer.

The way pigment affects vision is more complicated. Normally, when the irises are faced with glare they activate the pupils, a pair of gatekeepers that control how much light reaches the back of the eye. Without this regulation system, stray light enters through the pupil and iris, impairing the development of the retina and interfering with the optic nerve (the wiring system that connects the eyes to the brain). Albinism also affects the development of the fovea, a cluster of cones in the middle of the retina that are responsible for visual acuity. At around six weeks, almost every baby with albinism will develop nystagmus, in which the eyes dart back

and forth involuntarily. We don't know why this is a factor, but it is unrelated to pigment.

What we do know is that low pigmentation causes photophobia, meaning that daylight, particularly the searing rays of high noon, can be intolerable. It resembles those initial moments of squinty-eyed discomfort the rest of us feel when exiting a dark theatre into the light of day. Together, this complicated cocktail of eyesight issues is called low vision, and it is like seeing the world through your iPhone's Hipstamatic lens. The pixels are bigger, the world is a little brighter, and while it is not blurry the finer details are lost.

There are few experts in this field. The condition falls across a spectrum of medical specialties—genetics, ophthalmology, dermatology—and most general practitioners will never see a patient with albinism during their careers. When we visit our family doctor a week after we are discharged from the hospital, she dismisses my husband's concern about the janitor's comments. "I've seen babies this fair before," she tells us. Her file notes from our visit on January 5 list that Sadie has very fair skin, that her eyes are normal, and that she is thriving. Thriving! My maternal pride swells. My baby is flourishing. My husband, however, is not doing well at all.

Hours after our doctor's appointment, we take the dog for a walk and stop at a nearby schoolyard, where he runs in circles, chasing his doggy shadow across the floodlit snow. Sadie is tucked into her dad's coat, strung up in a contraption that keeps her close to his chest and out of the cold. We shuffle silently back and forth to keep warm. A heavy darkness fills the air between us. It followed us here, stalked us down the stairs from our apartment and along the night streets. It has been with us since we left the hospital. There is something wrong with Andrew. I will find him sitting quietly in the dimness of early evening. I switch the lights on, and he turns them off again. Even his camera, a constant flashing light from Sadie's first cry, has gone dark. But when I ask him what is wrong, he cannot find words to tell me.

My husband is the kind of person who leads the pack in a crisis. This strength, along with his height, his dark hair and green eyes, and his ridiculousness, are what had me pining to be his sidekick. But since the birth of our first child, he has come loose. He is distant, and unreachable. Parenthood exposes his Achilles heel, shocking both of us. What I don't know is that, like his parents, Andrew is convinced our newborn baby girl has a rare genetic condition.

My in-laws arrive the next day. Don carefully examines Sadie, using the contents of his doctor's tool kit, a ritual I wrongly assume follows the birth of every grandchild. He takes on the responsibility that, as a grandfather, he had hoped to avoid. He waits until morning, when his son, so clearly tormented, comes to him. Andrew is saddened, but receptive to the possibility of a problem. Sadie sleeps in my arms while he relays his father's concerns, releasing his own bottled fear in the process. To me, the suggestion is infuriating and impossible.

When noon comes and I still have not contacted my parents, it is gently suggested that I make the call. Sitting in a rocking chair by the nursery window, phone in hand, I stare out at the familiar scene and find it distorted. The row houses, stacked one above the other up Prescott hill, the two towers of the St. John's basilica, the grey winter sky—it is all askew. I dial and wait for my mother to answer.

"There might be something wrong with Sadie," I tell her.

There is a catch in my throat, and I can't continue.

My mother, listening on the other end, does not hesitate.

"No one will love her any less."

I do not fail to notice the peculiarities of my daughter's arrival, but I interpret them in a completely different way. My husband is a biologist, attuned to the natural order of the world. I am a folklorist, and walking the line between fantasy and reality is my work. I believe in science, but I understand fairy tales. My new baby's astonishing white hair and unusual beauty, her immediate legion of admirers, even the timing of her arrival—a

labour that stretched across some of the holiest days of the liturgical calendar—have the markings of a supernatural tale.

We mythologize even our routine birth stories. The most extraordinary reside in the world's grand narratives, from ancient Greece to the foundations of Christianity. Like the detailed version of Noah's birth, brought to public attention in the 1940s with the discovery of the Dead Sea scrolls. In it, the boy is born with flesh as white as snow, hair as white as wool, and unusual eyes that illuminate the room. His father, Lamech, is disturbed by his newborn son's appearance, so different from his own. He is suspicious, too. Recently, there were rumours that angels had been cavorting about with mortal women, and this child has definite angelic qualities. He consults his father, Methuselah, who in turn seeks the counsel of his father, Enoch. What Lamech ultimately discovers is that the white hair, luminous eyes, and pale flesh are attributes of the child's divine calling. "Call his name Noah," Enoch advises. "When all mankind who are on earth shall die, he shall be safe."

When texts from some of the scrolls are published in the mid-'50s, this birth story catches the attention of a British ophthalmologist named Arnold Sorsby. In 1958, he publishes an article titled "Noah: An Albino" in the *British Medical Journal*. He writes that the narrative is "clearly not that of a miraculous child but of an albino." To help prove his point, he includes a genetic breakdown and an adjoining diagram explaining the possible inheritance pattern of Noah's albinism. Only in the final paragraph does he suggest that the article is a parody, when he earnestly considers the recessive genetics of angels.

I read this paper shortly after the birth of my own ethereal child ("Your baby looks like an angel!" exclaims another new mom at the hospital). I search for Sorsby but find an obituary rather than a white pages listing. What I glean from his life story is that he edited the *Journal of Medical Genetics* for seven years in the '60s, he was an ophthalmologist employed at London's Royal Eye Hospital, and he specialized in genetic conditions of the eyes. All of this posits him as a person whose theories you would be inclined to take seriously. Case in point: when the first American albinism advocacy group forms in the '80s, it takes the acronym NOAH (National Organization for Albinism and Hypopigmentation) as its official name.

I show Sorsby's article to Dr. Daniel Machiela, a professor in the religious studies department at McMaster University in Hamilton, Ontario, with special expertise in the interpretation of the Dead Sea scrolls. He is interested but unconvinced.

"There is a metaphorical and symbolic attachment to the way he looks," Machiela says. "And that clearly seems to be what is going on here."

I want to connect Noah's story to my own, so I suggest that his ancient Near Eastern parents theoretically would have had dark hair, skin, and eyes, and therefore a child born with white hair would be very unusual.

"The point in these stories is that he was not just like anyone else who was born then—the way you would expect them to be born," says Machiela. "He stood out."

When Sadie is five weeks old, we meet with a geneticist, Dr. Lesley Turner. She is exquisitely gentle while examining our infant daughter, and I trust her immediately. We have seen an ophthalmologist, and we understand that Sadie has characteristics of albinism, but the doctor refers us to the Provincial Medical Genetics Program for conclusive tests. Andrew and I sit at a round table in an office at the Health Science Centre in St. John's, and I nurse Sadie while Dr. Turner and a genetic counsellor draw our family tree—a narrative of various disasters that includes an uncle who died too young of MS, a brother who died even younger of alcoholism, and on both sides the shattering experience of Alzheimer's.

Sadie has five millilitres of blood taken, half the regular amount, because she weighs just eleven pounds. She is silent when the needle pierces her skin, but she pees from the shock of it. The tiny vial of blood is flown to the University of Minnesota Physicians Outreach Laboratories, where I imagine a flurry of strangers in white lab coats carrying beakers and punching codes into complicated machines. The results arrive four weeks later:

Sadie has oculocutaneous albinism Type 1 (OCA1) variants a and b.

In OCA1a, the enzyme tyrosinase, which converts the amino acid tyrosine into melanin, fails to carry out its assigned task. In OCA1b cases, it makes a partial effort, and there is some pigment formation: yellower hair and eyelashes, darker eyes. OCA1 occurs with one in every 40,000 births. The recessive gene can be passed on silently for centuries, because both parents must be carriers for the condition to manifest. It is so rare, so improbable. Of all the gin joints in all the towns in all the world, Andrew walks into the Ship Pub in St. John's on a blustery June night. I spot him across the bar and think he looks familiar, so I introduce myself. The rest is genetic history.

It is a strange relief to succumb to your DNA. Earlier that week, I had fought back tears when a worried nurse at a lactation support session looked into Sadie's eyes and asked, "Does she smile at you? Does she make eye contact? Can she focus on an object?" No. No. And no. But with the albinism diagnosis, I throw out all of my "baby's first year" books and ignore the monthly milestones attributed to normal development. The first time Sadie reaches for an object (a garish purple dragon hanging from the handle of her bucket seat), the first time she holds my gaze, the first time she smiles back at me, these will happen on a different timeline, and they will be some of the most exciting, profound moments of my life.

When I meet with Dr. Turner a year later, I ask her how it feels to be a genetic code messenger. She considers this for a moment. In our case, she has noticed a shift toward acceptance since our first visit, particularly in me. Andrew, his earlier depression lifted, seems receptive after the initial meeting.

The hardest cases are when a child's prognosis is terminal. She tells me about walking into the small room where we met the previous year and facing an entire family (child, parents, and both sets of grandparents) to deliver the news of the fatal genetic flaw. The mood was heavy. The father was weeping. Dr. Turner excused herself for a moment on the pretense of finding a few more chairs. Instead, she went into her office, put her head down, took a few deep breaths, and said, "Okay, pull yourself together."

"Then what?" I ask.

"And then I was fine to go back in," she says.

"It's not supposed to look like this," says a fierce, sad-eyed mother in the outpatient waiting room at the children's hospital in St. John's. "You get pregnant, and you have a baby, right?" She shakes her head. "I had a big bleed at thirty weeks, and it's been hell ever since." I have seen this mother before; I take Sadie to a slew of specialists with offices here, and our appointments often coincide. The woman points out her daughter, a small eighteen-month-old with a wiry build and corkscrew curls seated at a miniature yellow table, colouring with conviction. I don't see the problem, but it turns out that no one does. She suffers from a failure to thrive. This is the term allotted to cases where an infant or a young child stops developing at the same rate as her peers. They begin the regular way, and then there is a change.

In European fairy lore, a newborn whose nature turns foul, who screams with colic or falls suspiciously quiet, whose chubby cheeks turn gaunt and whose bright eyes hollow, is called a changeling. It is sometimes believed that the human child is switched by the fairies for one of their own offspring. The fairies persisted in North America, stealing infants from cribs throughout Newfoundland and leaving cross, wizened, unfamiliar babies in their places.

Some scholars attribute changeling narratives to cases of failure to thrive. A supernatural explanation absolves the parents of guilt; rather than a genetic hand-me-down, it is a case of switched identity. It allows them to grieve for the stolen child, the one they had conjured over nine long months—because that child is gone, away with the fairies, and with it any preconceived ideas of parenthood.

There are no changeling stories connected to albinism because, with the exception of nystagmus, the

condition does not develop after birth. However, the ethereal whiteness does inspire albinism lore across the globe, particularly where people typically have darker skin, hair, and eyes. In the early twentieth century, New Zealand ethnographer Makereti Papakura found that Maori people with albinism are believed to be the offspring of mortal women and supernatural men who belong to a tribe of fair-haired mist dwellers.

Around the same time, Western scholars sought to verify rumours of an “albinotic race” residing among the Cuna people of the San Blas Islands, off the coast of Panama. The theory has been debunked, but the place still appears to have a high incidence of the genetic condition. D.B. Stout, an intrepid anthropologist, takes up residence among the Cuna and discovers that in some communities, individuals with albinism are associated with higher intelligence, godliness, and magic powers that enable them to ward off a demon that periodically eclipses the sun and moon. Stout’s successors find that Cuna people with albinism are called moon children, alluding to their mothers or fathers staring too long at the night sky during gestation.

I print the albinism lore articles and keep them in a folder on my desk. They sit side by side with Sadie’s five-section medical binder. In some ways, what I learn about the moon children of the San Blas Islands is just as important to my understanding of the condition as the literature from our genetic counsellor.

In June, we take Sadie to see Dr. Elise Héon, the chief ophthalmologist at the Hospital for Sick Children in Toronto. She is a tall, handsome woman, wearing a sleeveless Anthropologie dress stamped with a bucolic landscape print. She graciously acknowledges the distance we have travelled and makes us feel like visiting royalty. She is a commanding presence, and her underlings are practically tripping on her white coattails. Even Sadie, traumatized by an earlier test involving electrodes plastered to her head and a strobe light, succumbs immediately to the doctor’s charm and reaches for her arms. In faintly accented but excellent English, Dr. Héon informs us that our daughter will never drive a car.

I can no more picture six-month-old Sadie driving a car than I can imagine her using a fork. The future is slippery and hard to grasp. Sometimes, I imagine approaching her teacher to discuss how to avoid flash photography on picture day. “Perhaps you would consider taking the pictures outside this year?” my future self will ask Sadie’s future teacher, explaining how the flash makes her blue eyes appear red. But when I visualize this, I am standing in the foyer of my own elementary school and speaking with Mrs. Vijendren, my grade two teacher. I have no frame of reference for Sadie’s future nor, for that matter, my own.

After our morning at SickKids, we walk over to the Art Gallery of Ontario to see the abstract expressionist show. In the hushed world of the exhibition space, my mind wanders. Sadie is asleep in her stroller, buried beneath the hulking grey UV shield. Several of Mark Rothko’s giant colour field paintings dominate a corner of the gallery, floating landscapes of shimmering hues, detail-less and yet so emotive. This is how Sadie will see, I think, in giant fields of colour, as an abstract expressionist does.

A few days later, I am cornered at a family gathering by a relative’s friend, a tireless raconteur of questionable narratives.

“My aunt is an albino,” she says. “She had to wear a blanket over her head during the day, if she ever went outside, but mostly they had her working in the fields at night during rainstorms. Everyone thought she was a witch.”

“Really?” I ask, unconvinced.

“You got the short end of the genetic stick,” she says, shaking her head.

It enrages me that this homely scale tipper is calling my beautiful baby a manifestation of poor genes. But it is the interactions during the most mundane aspects of life that wear me down. A flash mob of shoppers comes alive at the grocery store when I push my daughter along the aisles in the cart—craned necks, pointed fingers, wide-eyed astonishment. “Does she get her hair colour from you or your husband?” they ask. The answer is

both, of course, because it's a recessive trait.

Sadie gets fitted for glasses in the fall, to help control her nystagmus and improve her distance vision. Something about the pink plastic frames digging into her chubby cheeks upsets me deeply. I erupt in great, galloping sobs at the optometrist's office. Even after I pull myself together, a snivel will escape at random, like a hiccup.

Around the same time, I am roused from sleep one night by a drunk abusing the cars parked on our downtown street. His fists repeatedly rain down on hoods and trunks and windshields. The bashing of flesh on metal and glass sounds like an offbeat drum, punctuated by his raging diatribe. The next morning, I see two cars with smashed windshields on our block. Later that week, a neighbour tells me that the offender returned, leaving an envelope with \$250 on the windshield of one of the damaged vehicles. Inside, a note read, "I'm sorry I smashed your windows. I don't know what got into me."

This outburst and the man's apology resonate with me. Some days, I want to scream out my front door like a banshee and smash my world apart, then leave a sorry note and get on with life.

Instead, I rub two shiny worry spots into either side of my scalp. While organizing photos from a spring vacation, I am shocked to see the round bald spots peering out of my hairline, like a second set of eyes. Motherhood, with its saggy-bodied sleeplessness and the baby's constant cry, is a foreign country. Not Tibet exactly, but certainly Denmark, or Croatia.

At eight months, Sadie begins to spend time at our local CNIB (founded as the Canadian National Institute for the Blind), a centre where we can use a playroom designed for children with limited sight. It is called the Snoezelen Room (an awkward Dutch neologism that pairs the words for "sniff" and "doze"), and it conjures hazy memories of European discotheques from the year I spent in the south of France. A sometimes darkened lair, it is a wild array of flashing lights, mirrors, padded floors, lit-up toys, and beanbag chairs, under a convincing projection of stars. There is another projection of an underwater scene in the shape of a circle on the wall, as if we are peering out of a submarine porthole. Sadie's vision progresses with each visit, until one day she notices the stars on the ceiling.

Along with the music lessons, playgroups, and kids' gym, the Snoezelen Room becomes just another new mom-and-baby excursion. Months back, however, pushing my stroller through CNIB's doors for the first time, I agreed with the woman I met in the hospital waiting room in St. John's: "It's not supposed to look like this."

But it does. And it will. I see lots of other kids wearing sunglasses, and we live in an age of UV-proof clothing and SPF awareness. Visually impaired or sighted, we all carry technological devices that facilitate our everyday tasks. Sadie is beautiful and smart and ridiculously funny, and most importantly she is loved. Her network starts with her two smitten parents and expands across family and friends, a team of doctors, and a beloved dog that waits with tail-thumping enthusiasm at the nursery door every morning. Her fans include the Ph.D.-wielding mamas in our baby group, her sitters, the besotted employee at our local grocery store, and our postal worker, who for a year delivers weekly packages to the little blond girl at number sixty-two.

Before Sadie came along, smug parents would tell me that you can only really know love when you have a child. I interpreted this to mean the love you feel for your child, which I now know is vast and indefinable. But I wonder if they meant it in a greater sense. It is the love we receive that astounds me. You never know how much people care about you until you fall apart a little and everyone picks you up, piece by piece, and puts you back together again.

I am the anthologist, and these are the stories in my repertoire. Occasionally, snippets of darker narratives sidle their way into my collection. I read about folk beliefs in Zimbabwe and Tanzania, where some see albinism as a curse, a contagion, or punishment for a mother's infidelity with a malevolent spirit. In these nations,

decimated by disease, there are rumours that sleeping with a woman with albinism can cure HIV/AIDS.

“The same culture that can elevate me to a god can turn me into a demon,” says Peter Ash, a British Columbia resident with albinism who founded Under the Same Sun, a Vancouver non-profit that strives to better the lives of people with albinism in Tanzania.

Ash is a fast talker, and he rhymes off the atrocities visited on Tanzania’s albinism population in rapid-fire succession: rape, violence, dismemberment, social stigma, abandonment, orphanhood, and infanticide. He tells me about visiting a little boy who had lost a hand to poachers—a transaction set up by his parents. The poachers sell “albino” body parts to witch doctors, who believe they hold magical properties and use them for curative potions.

Ash remains remarkably objective when discussing these issues. I am less so. Whispers of colonial judgment shroud my objectivity. “How do I interpret this?” I ask my former teacher Diane Goldstein, a folklore professor at Indiana University, and a renowned belief scholar who describes herself as a “cultural relativist.” She reminds me to look at the context, and says that in her own work (on AIDS legends and beliefs and, more recently, infanticide), she finds that “at the heart of these beliefs are important cultural issues that are very humane.”

And how different are these beliefs from our own? Hollywood is North America’s witch doctor, and just as barbaric. Such popular films as *The Da Vinci Code*, *The Matrix Reloaded*, and *The Princess Bride* all feature albino villains. Despite our perceived modernity, much of our faith and knowledge is wrapped up in make-believe.

After months of reading parenting memoirs and medical journals, I return to novels. I pick up Michael Crummey’s *Galore*, and in the first few pages a “bleached white” man is cut from the belly of a beached whale, to the horror and fascination of the outport villagers who witness the event. The man is christened Judah, and there is a hint of the supernatural in his muteness and whiteness. He is often referred to as “the albino.”

I have crossed paths with Crummey a few times over my years in Newfoundland. He is kind and affable, and I like how he peppers his soft-spoken sentences with emphatic curses. He agrees to speak with me about Judah’s character.

We meet in a soulless boardroom in the library at Memorial University—Crummey’s alma mater, where I am working on my Ph.D.—and he tells me that Judah’s whiteness is not related to a genetic condition. (In the book, a doctor’s examination reveals that Judah is “not a true albino,” and Crummey jokes, “I guess given your own experience he could have just got it wrong.”) The decision was practical: Judah was bleached by the whale’s stomach acid. However, the whiteness opened up a variety of literary possibilities.

“White is the empty page,” says Crummey. “It’s the blank canvas, and so what I saw happening in the book was that Judah was the blank canvas people could project whatever they desired most—or whatever they feared most—onto. The community created Judah over and over and over again because he was blank.”

White is the non-colour, both enigmatic and profound. I think of Noah’s whiteness, symbolic of a divine path. I think of the fair-haired people who live in the New Zealand mist, the moon children on a string of islands off Panama, and the infants who went away with the fairies of Newfoundland. I think of the tiny white-haired baby who slept in her plastic bassinet while half of the hospital came to her side in awe.

I can’t go back to those few days in the maternity ward, before the janitor’s Cassandra-like prediction, before science threatened my version of the story—when I spun my own tale and accepted my daughter’s beauty as otherworldly and magical. I am not certain that we are better off for knowing the molecular story rather than the folk tale, or whether there is room for both. Science can tell you how genetic anomalies and birth defects happen, but not why they happened to you rather than your neighbour. Medical facts can rarely offer the level of comfort that stories can. At least in our personal narratives, we have control. Here is the value of folklore: it

gives shape to the unknowable. This can be uplifting or dangerous, but ultimately it explains human difference in a way that science never will. Some days, I yearn for my short-lived dark age, but it is a curious nostalgia, because I can't imagine life—and more specifically my daughter—to be any different, and in the end I don't want to.

When Sadie is a year old, Dr. Turner invites us to share our tale of genetic discovery with her first-year medical students. We nervously over-prepare with a twenty-one-slide PowerPoint presentation and seven pages of notes.

The talk goes smoothly, until we broach the topic of having a second child. There is a one-in-four chance of albinism, and one in two that the baby will be a carrier regardless. Then there is a one-in-four chance that the gene will not be present at all. These numbers tell me as much as tea leaves or tarot cards. A second child will have this condition, or she will not.

For now, it is just a line in our printed notes: "Talk about DNA/in vitro testing." The rest of the sheet is blank, because we have come to the end of the story so far. We have to look out at the fifty or so faces in the room and shrug our shoulders, helpless to fate, helpless to a wiring system I barely understand, the invisible ruler we must obey that hovers, depending on what you believe, somewhere between God, fairy tales, and science. The talk stops here. The students clap, and Sadie looks up from the third row, where she has wedged her walker between a desk and a knapsack. The clapping ends, and the students ask us questions we can confidently answer, about events that have already happened. Sadie dislodges her walker, turns, and races off in the other direction, happy to receive the students' attention, content to hear her parents' voices in the background, knowing that we are there, watching her and making sure (for now) that her world is padded and safe.

Telling our story to the medical students is not as frightening as I had thought it would be. The reality is that I tell versions of this story every day. I tell inquisitive grocery shoppers, and moms at playgroups; I tell my seatmates on airplanes, and strangers at the park. I perform the narrative like a folk tale, many times, and it changes depending on the context and the audience. One day, I will pass it on to the person who matters most, because it is her story, after all. I wonder how she will tell it.

Related Links

["My Dad, the Movie, and Me"](#) by Noah Richler (*October 2010*) • On the set of *Barney's Version*, with Dustin Hoffman, Paul Giamatti, Minnie Driver, and the ghost of Mordecai Richler

["The Changeling"](#) by Gail Gallant (*July/August 2006*) • On top of the TV there was a picture, colourized and framed, of baby Gail sitting on my father's knee, with her name printed in the top right corner: Gail Gallant at 3 months

["Cause and Effect"](#) by Lynn Cunningham (*October 2009*) • Fetal alcohol syndrome is preventable, incurable, and surprisingly common. Still, it brought me Boop, who has redefined my life

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Byron Eggenschwiler has published work in the *New York Times*, the *Wall Street Journal*, *Quill & Quire*, *Businessweek*, and the *National Post*.





“SLANG”—Sensitive Language and the New Genetics—an Exploratory Study

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Published Online: 29 December 2005

SLANG—Sensitive Language and the New Genetics—is a concept that arose out of informal discussions between a number of interested parties, both consumers and professionals, who were becoming increasingly uneasy with some of the language commonly used in medical genetics. Some language choices were felt by the authors to be inappropriate for a variety of reasons. Poor language choice may impede an individual’s understanding of a genetic condition or important medical information and the chosen words themselves may simply be perceived as discriminatory and even offensive.

SLANG is an important concept to explore partly because literature in this area confirms that language choices in medical settings can be of great significance to both patients and families. Studies have shown how language choices impact on professional practice by, as one example, changing the intended meaning of medical information and affecting individual perception of risk and choice which, in turn, may affect individual or familial well-being. In addition language choice has the power to affect how individuals perceive themselves and are viewed by others. This paper presents the results from our pilot study and discusses the implications for health professionals with particular reference to medical genetics settings.

KEY WORDS: consumer; language; health professional; medical genetics.

INTRODUCTION

The concept of SLANG—Sensitive Language and the New Genetics—came from discussions between genetic health professionals and consumers in Melbourne, Australia in response to concerns about

some language choices that were felt to be inappropriate or confusing.

Clear and effective communication should be of paramount importance to those working in the field of medical genetics. Sociolinguists recognise the power of language choice and suggest that words have the ability to influence individual and community perceptions of “reality” (Holmes, 2001, p. 331). For health professionals it becomes particularly prudent to make wise word choices, given the position of power and responsibility in which doctors often find themselves (Grbich, 1999). It has also been recognised that health professionals are particularly prone to using “demeaning shorthand or jargon that stereotypes or dehumanises” (Rothstein, 1991, p. 498). Language choice should be seen as a key discussion issue in health care generally and the exploratory

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study outlined in this paper highlights the need for language choice to be a particular priority for those working in medical genetics.

Language choice has been shown to affect a number of different areas that are particularly relevant to the genetic counseling process.

Framing of Genetic Risks

Client perceptions of risk may be affected by a number of ways in which the risks are framed (Shiloh and Sagi, 1989; Abramsky and Fletcher, 2002; Sarangi *et al.*, 2003) and this may have an effect on risk processing.

For example individuals may perceive a risk that is presented in the form of a percentage (e.g. 10%) as either higher or lower than the same risk when it is presented as a proportion (e.g. 1 in 10) depending upon how that individual reasons numerically (Kessler and Levine, 1987).

Client Anxiety Levels

Word choices in genetic contexts can play a part in both reducing and increasing guilt that is sometimes experienced by parents of children with a disability (Chapple *et al.*, 1995).

Anxiety levels may be increased when professionals use genetic terms with names that are unfamiliar and may “conjure up alarming images” (Chapple *et al.*, 1997; Gordon *et al.*, 2002). In one study the word “mutation” for example was found to have negative connotations for lay people and the authors suggest that health professionals should avoid using that word when talking to families about genetics (Condit *et al.*, 2004).

Autonomy

A nondirective counseling style is generally valued by genetic counselors as a means for allowing clients to make their own choices i.e. respecting client autonomy. Few words are actually value free and it is possible that some language choices may be perceived by clients as directive, for example in prenatal settings referring to a “baby” rather than a “fetus” while decisions are pending about prenatal testing (de Crespigny *et al.*, 1999).

The manner of speaking may also impact on a client’s ability to make autonomous choices. Discourse analysis has demonstrated how genetic coun-

selors may use “indirect speech” (e.g. use of “some people,” “most people”) as a way of trying to maintain neutrality and how this may be in conflict with attempts to explore clients individual needs (Benkendorf *et al.*, 2001).

Respect for Persons

Effective medical information communication needs to be both relevant for and inoffensive to the target group. Culturally sensitive communication materials have been developed specifically for African Americans with a family history of cancer (Baty *et al.*, 2003). Ideally sensitive language should be an integral part of medical communication but this is more obvious in certain areas than in others.

1983 was the International Year of the Disabled and since that time “Person-first language” has been developed (Titchkosky, 2001). It came from the recognition that language has the power to offend and degrade individuals as well as influence perceptions of disability in society resulting in increased stigma and discrimination. Preferred terms include using “person with a disability” in place of “handicap” and “persons who are blind” in place of “the blind.”

Setting for Research

The Murdoch Children’s Research Institute (MCRI), Melbourne, Australia, is a multidisciplinary research facility incorporating laboratories, clinical genetics, ethics, education and public health divisions. In addition it is host to the peak consumer organisation Genetic Support Network Victoria, and as such it is an ideal environment within which to explore the concept of SLANG.

METHODS

At a state-wide conference for genetic lay advocacy and support organisations in Victoria, Australia, participants were invited to offer their opinions on some of the language that is frequently used in medical genetics settings. The one page questionnaire nominated seven groups of words and asked participants to indicate which words they found offensive or unsuitable. Consumer and professional discussion surrounding particular areas of language experienced as problematic informed the lists of words used. The words and phrases were grouped accord-

Table I. xxxxx

Words ^a	Percentage of respondents who find find word offensive	Alternatives offered
Abnormality	56	Birth condition difference
Birth defect	47	
Birth anomaly	34	
Congenital disability	22	
Genetic disease	53	
Genetic disorder	19	
Genetic condition	3	
Mental retardation	81	
Slow learner	44	
Intellectual disability	13	
Developmental delay	9	
Learning difficulty	3	
Mutant gene	44	
Defective gene	34	
Faulty gene	28	
Allele	22	
Mutation	22	
Gene change	6	
Spastic	97	Person affected by...
Retarded	94	
Handicapped person	69	
Disabled person	38	
Person with a disability	0	
Person with special needs	0	Apparently normal average usual
Normal	28	
Unaffected	13	
Foetus	19	
Embryo	19	
Baby	6	
Pregnancy	6	
Unborn baby	3	

^aWords highlighted in bold were considered offensive by a majority of respondents.

ing to the contexts in which they might be used (see Table I), and did not necessarily have the same meaning. Respondents were also invited to offer alternatives to the words listed. The final section of the questionnaire comprised open-ended questions asking for general comments on language. The thirty-two respondents comprised people with genetic conditions and their advocates.

RESULTS

Analysis

Analysis of the results was simply done by counting the number of times a response was given to a particular question. From the comments we

were able to extrapolate a number of recurring themes.

Themes Arising from Comments Made in the Study

• **Not Normal...**

It was commented by respondents that everyone is normal. Alternatives offered were

“apparently normal” and “average”.

Someone else suggested “usual”:

I use ‘usual’ when talking about sex chromosome issues with XO or XY girls. ‘XX is the chromosome arrangement usually found in girls, but there are exceptions’—

thus establishing that the patient's a girl, not something else.

Another respondent commenting on context said,

If words are not in context it is hard to make a judgement i.e abnormal used objectively or personally?

- **Use of the term 'disease'**

It was commented by one respondent that they:

Generally prefer 'condition' rather than 'disease'.

Another person said:

All people, but especially doctors, need to use the term disorder and not disease. Disease is transferable to other people—contagious—passed on by germs. A disorder is a dis-order within our bodies—outside the normal.

It has previously been suggested by others that words or phrases such as “medical condition” “birth variation” “person affected by . . .” (Briffa, 2002) are preferable to the term ‘disease’.

- **Frightening terms**

While a “gene mutation” may well be strictly correct as a cause for disease in scientific terms, its meaning is different in common language, as recognised by this comment:

Mutant is tainted from space alien movies—best avoided except to say it means ‘changed’.

Another alternative, which was offered, was “*altered gene*”.

It was commented that the term “allele” could be too technical to use in this context.

- **Person First language**

The comment was made that,

In general ‘a person with’ is preferable to ‘he is a’

A similar comment was:

I think it's good to use ‘people with . . .’, ‘people affected by . . .’

and

avoid, where possible, label nouns e.g patients, sufferers, victims and words ending with ‘ic’, like epileptics etc.

- **Suggestions given by respondents for professionals**

Avoid medical jargon.

Important for professionals to be aware of non-verbal - tone of voice, body language. Need to be self aware – do they feel uncomfortable, awkward using various words.

One of the biggest challenges is to avoid language which conjures up thoughts of parental guilt/blame or divine punishment.

Avoid terms which make people feel bad about themselves or their children. ‘Retarded’ is one of these words, ‘normal’ is another. The Short Statured People of Australia taught us to talk about ‘average’ or ‘usual’ stature instead of ‘normal’.

DISCUSSION

This preliminary, exploratory study confirms our belief that SLANG is an important concept worthy of further exploration.

The survey coupled with the initial discussions that the authors have had with consumers and health professionals highlight the need for promotion of a universally acceptable language which may ease the perceived tension between medical personnel and consumers as well as facilitate better client education.

SLANG recognises that over time the meanings of words will change (Holmes, 2001), and what may have started out as a purely medical description may eventually gain additional, and sometimes offensive meanings. One example of this can be seen in the words once chosen to describe Down syndrome. When the condition was first recognised in the late 1800s those living with Down syndrome were frequently referred to as idiots, imbeciles or Mongols. John Langdon Down himself wrote . . .

“a very large number of congenital idiots are typical Mongols” (Down, 1887 quoted in Ward, 1999, p. 20).

Other evidence of how acceptable language changes over time was shown by The Spastic Society of Victoria who as a result of perceived negativity about the name it had held for 53 years became

SCOPE. They felt that the new name conferred a more positive identity as the term ‘spastic’ had developed over time into an insult.

While it is important to acknowledge individual language preferences we must also recognise that there are some words, which may now be universally unacceptable.

Discomfort with the use of stigmatising labels was one of the primary reasons for the development of SLANG. We are not suggesting that it is possible to create a language that could be devoid of problems or misuse: *Rather we seek to raise the awareness of health professionals on the importance of language choice.*

Aside from our respondents others have recognised that

“words and the images they convey can set a tone for social acceptability” (Rothstein, 1999, p. 498).

Future exploration of SLANG will aim to ensure that geneticists, genetic counselors and others working in the field of medical genetics do not remain in a linguistic time warp. In addition discussion may promote flexibility and responsiveness in language choices, while recognising that what one person may find acceptable another may not. This was clearly demonstrated in an on-line survey conducted by the British Medical Journal exploring health-related words that people would like to see banned. It discovered that 18 respondents wanted the term “health professional” banned, 77 wanted to get rid of the term “clients”—in preference to patients, and 10 people objected to the term ‘doctor’ (bmj.com, 2002).

Our discussions with health professionals in Melbourne highlighted that in practice it may be difficult to be both technically accurate and sensitive. While we acknowledge this position we feel that it should be possible to find local consensus in regard to the most commonly used terms. We accept that at times it may be difficult for health practitioners to strike a balance, but it is precisely this difficulty that makes it vital for professionals to be reflective and reflexive about their practice. Nowhere is this balancing act more evident than in the discussion of medical “risk”, a common concept in medical genetics. For some it may be that the very concept of risk is inherently negative (Linell *et al.*, 2002). How for instance, can one talk about the risks of a patient’s child being born with a medical condition without making that “risk” sound somehow menacing or losing the medical or scientific facts amongst the background “chatter”?

Could more sensitive word choice be the answer? Words such as *chance*, *prospect*, *probability* and *likelihood* may, for some, have less negative connotations attached than *risk*, and could be used without jeopardizing the scientific or medical validity of what may need to be communicated.

Another example exists in the area of prenatal testing where it may be reported that a pregnant woman “*refused*” amniocentesis. If, as we hope, an offer of prenatal testing is really a choice then it would be more correct to say that a pregnant women “*declined*” amniocentesis as that term better reflects the idea of choosing between two equal options.

Again SLANG contends that there may not be any hard and fast rules, rather we urge practitioners to consider the available choices.

Importance of Consumer Consultation

The long-standing historical model of health services as charity has defined the way services have been provided until recent decades. With a shift towards consumer empowerment in many areas, have come the beginnings of consumer consultation in the planning of health services. Reasons given for inclusion of consumers in planning services include the chance to improve the quality of services and attain accreditation in quality improvement schemes; improving health outcomes for patients; providing a more responsive service and therefore improving uptake and observance rates; and most fundamentally, because equal participation is considered an ethical right in democratic societies (Bastian, 1998; Allsop *et al.*, 2004). This final concept is under-pinned by mutual respect between consumers and practitioners, and the authors suggest that a strong relationship with consumers and their representative organisations will contribute to the skills and insight of practitioners.

As demonstrated by the results from the pilot study, consumers do have a number of useful suggestions for health professionals—many of which are dependent on the professional being reflexive, insightful and empathic.

In addition poor language choice is a major contributor to the plethora of medico-legal cases in the court systems of countries such as the United States (The Back Letter, 1994). We believe that careful choice of words may reduce the risk of labelling and stigmatisation, and prevent unnecessary consumer anxiety.

Limitations to Study

There are obvious limitations to this pilot study. This was a small exploratory study not intended to be generalisable but rather to investigate whether language was as important for members of lay advocacy groups as we had imagined.

Again we acknowledge that language use will vary significantly between countries and even within institutions, any suggestions given in this paper, are just those... suggestions.

What this study and the authors strongly recommend is further research⁶ into the area of word choice and genetics, as well as extensive exploration into any impact language choice has on consumers and their families. Using direct questions we were able to explore the impact of certain words for this group of consumers, other methods such as empirical message effects studies, focus groups and semi-structured interviews could provide a broader research base. In addition input from the lay public and health professionals as well as new consumers would enrich the debate and more effectively contribute to development of evidence based best practice.

CONCLUSION

This study has allowed us to demonstrate the relevance of language choice for both consumers and health professionals in our small cohort and has enabled us to recognise the importance of further studies in this area.

Our vision is to prompt inclusive debate and encourage further research. We hope that this work will promote ongoing discussion and reflection that can become an integral and valued part of practice for all involved in medical genetics.

As Rudyard Kipling once said "words are the most powerful drug known to man".

ACKNOWLEDGMENTS

The authors would like to thank members of the Genetic Support Network Victoria (GSNV), Caroline Bowditch and our colleagues at Genetic Health

Services Victoria (GHSV) and The Murdoch Children's Research Institute (MCRI) for providing the impetus and opportunity for the development of SLANG.

We would also like to thank the reviewers of this paper for their helpful comments.

Jan Hodgson is currently undertaking a PhD and would like to thank The Gene CRC—Cooperative Research Centre for Discovery of Genes for Common Human Diseases, Richmond, Victoria, Australia for their continuing financial support.

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