SECOND OPINION: DISABILITIES: BABY KAYLEE'S SURVIVAL POINTS TO ISSUE

Rare diseases deserve special attention

Canada is one of the only countries without a plan to support people with 'orphan' disorders

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Earlier this month, the nation was captivated by the story of Kaylee Wallace after parents of the gravely ill infant decided to remove life support and donate her heart to another dying child.

But Baby Kaylee lived and the media moved on.

Now she is thriving, despite the challenges posed by Joubert syndrome, a brain malformation that leads to developmental delays, breathing difficulties and motor-skill problems.

Baby Kaylee is now one of millions of Canadians living with a disability, and one of the thousands upon thousands living with a rare disorder.

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Note here the repeated use of the verb "living." Many of the pathos-laden stories about the case implied that living with a disability was a fate worse than death.

This is nonsense, of course. Baby Kaylee's life, however different, can be as fulfilling and as wonderful as any other and it should be valued as much.

But this does not imply that it is easy to live with a severe disability or with a rare disorder. The indifference to the child once she was shifted from the "potential heart donor" column to the "disabled child" column is symbolic of society's attitude.

The media coverage of Baby Kaylee focused on her parents' heart-wrenching dilemma about removing her from life support. But the reality is that they face a far more heartbreaking plight in the struggle to help their child live a full and rewarding life.

If anything, Baby Kaylee's predicament should open our eyes to the gaping policy void that exists for the legions of children and adults who face similar challenges.

Canada is one of the only developed countries without a plan to support people with rare disorders. Heck, in Canada, we don't even have something as elemental as an official definition of rare disorders on the books.

Elsewhere, rare disorders are defined as those that affect fewer than one in every 2,000 people. There are an estimated 7,000 such diseases (and advances in genetics mean the number is growing).

All told, these "orphan" diseases affect close to three million Canadians to varying degrees. But unlike sufferers of common illnesses such as cancer and heart disease, they remain largely invisible and voiceless.

Rare or orphan diseases pose a lot of challenges to individuals, health professionals and the larger health system, notably:

Screening for genetic conditions is limited and varies from province to province, so children often go undiagnosed;

Because these conditions are rare, physicians may misdiagnose and mistreat rare conditions;

There is little financial incentive to produce drugs to treat conditions that affect only dozens or hundreds of people nationwide, and when there are drugs they tend to be expensive;

When drugs and other treatments (ranging from special food supplements to devices) are available, coverage can vary markedly from province to province;

Caring for a child with a chronic disorder or severe disability can be a full-time job, but parents get little support.

There are, thankfully, ready solutions to all these challenges:

You cannot realistically screen for every rare condition, but it is possible to focus on those that can be treated. Right now, provinces screen newborns for up to 30 genetic conditions, which is reasonable. But there is a grave shortage of genetic counsellors to help families understand what the findings mean;

It is not realistic to expect every physician to have expertise treating orphan diseases, but you can, like Europe, create referral centres where health professionals and families can go for specialized help;

The United States has an Orphan Drug Act that provides a series of financial and other incentives to encourage research and development of drugs for rare conditions. Canada is a laggard in this area and needs to catch up with a bold action such as creating an institute for rare disorders within the Canadian Institutes of Health Research;

Governments have, for years, talked about providing catastrophic drug coverage, an insurance program for those burdened by drug costs, and this would be of great help to those with rare conditions.

Good health care consists of more than medical intervention. Social support is also essential. There are insurance programs, private and public, for people with common chronic conditions, and these need to be adapted and bolstered for those with rare conditions.

The underlying philosophy here is that rare disorders, by their very nature, deserve special treatment.

Over the next two days, the Canadian Organization for Rare Disorders will be holding a conference in the shadow of Parliament Hill that highlights these and related issues.

Hopefully, participants in the Ottawa event will also take the opportunity to remind MPs that just last year, they unanimously adopted private members motion M-426 that called on the government to "respond specifically to the challenges faced by Canadians with rare diseases and disorders."

Since then, nothing has happened. Baby Kaylee and others in her situation continue to be orphans of medicare, and victims of shameful and heartless policy neglect.

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