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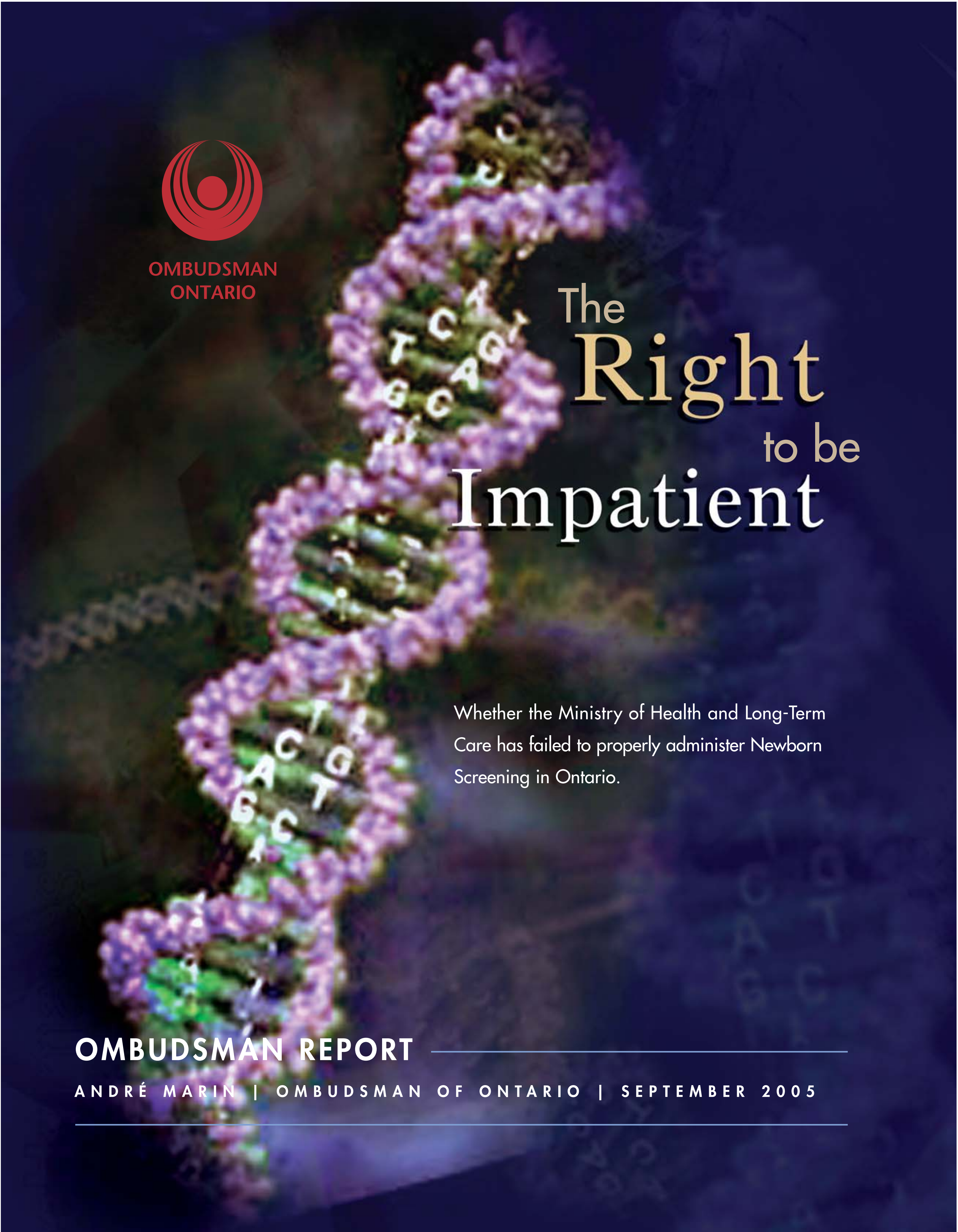

The Right to be Impatient

Whether the Ministry of Health and Long-Term Care has failed to properly administer Newborn Screening in Ontario.

OMBUDSMAN REPORT

ANDRÉ MARIN | OMBUDSMAN OF ONTARIO | SEPTEMBER 2005

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Ombudsman Report

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"The Right to be Impatient"

André Marin
Ombudsman of Ontario
September 23, 2005

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1 John Adams' child was lucky. Unlike dozens of other children who have died as a result of the failure in Ontario to identify detectable medical conditions, this child's rare disorder was discovered by newborn screening, something Mr. Adams gave thanks for in a recent letter to the editor.[1] In his letter Mr. Adams also commended the Government of Ontario for its recent announcement to increase from two to 21 the number of conditions that will be tested for. Yet as thankful as he no doubt is, this father was not motivated to write simply or even mainly to express his gratitude. He wrote to convey his deep concern that the Government of Ontario's decision could be a case of doing too little, too late, and too slowly. In his letter Mr. Adams cited statistics on the availability of newborn testing internationally, statistics that should embarrass all of us within this province. Once a world leader in newborn testing, Ontario has fallen behind to the point where, pending the implementation of announced changes, it continues as I have already publicly said, to perform like some "third world country."

2 How did this happen? I explore that question in detail in this Report but something general can be offered by way of introduction. Although not a technical explanation, Mr. Adams offered that the process of deciding what newborn screening should look like in Ontario is flawed – it failed for so long, he believes, because parents were not consulted and he suggests it will continue to fail if this is not changed. "There is a special expertise to be gained," he said, "from living night and day with a child with a rare disorder." In an electronic message to my Office Mr. Adams made that point eloquently: "Parents are the experts in the wasteful odyssey once a child begins to exhibit non-specific signs and symptoms before there is a diagnosis of a rare disorder not identified at birth."

3 For my part, I do not know whether a decision-making process on newborn screening does in fact require parental participation to succeed, but I do know this - John Adams was "bang on" in identifying the primary ingredient that has been missing in some Government quarters for the past decade or more that parental participation would have supplied, namely, the personification of misery. What was evidently missing for some key decision-makers was an appreciation that newborn testing is not just some impersonal, abstracted, bureaucratic question or an issue for another day. It is a matter about unnecessary illness, suffering and the death of real children. In spite of this, successive governments have responded as though their decisions or inaction carried no human consequence or aided and abetted no suffering. Over the past years our process of decision-

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[1] "Screening of newborns is expanding too slowly," The Ottawa Citizen, 14 Sep. 2005, A15.

making on newborn testing, if it can be called a process at all, was not the march of progress that we in Ontario have a right to expect. Instead, it was an ambling, staggering, distracted if not disinterested meander. It is therefore understandable that Mr. Adams would remark, now that a corner appears to have been turned and the Government of Ontario is promising to do better, “Parents have a right to be impatient.”

4 In this Report I do three things. First, I too commend the current Government of Ontario for finally shaking off the malaise that has made this province guilty by omission in the deaths and disability of dozens of our most vulnerable citizens. It is showing leadership in a Ministry that has failed on this issue for too long, and credit is due. Second, I chronicle the sad history of newborn screening in Ontario, in part for posterity but mostly for the lessons it can teach. It is better for us to perform a painful post-mortem on this bureaucratic lethargy rather than to sit idly by and wait for more unnecessary post-mortems on our children. But let me be clear. This report is not about recriminations and blaming. It is a cautionary-tale with an eye to today and tomorrow – one whose lessons can be used to make sure the current Government now moves resolutely as it has promised to do.

The Inspiration for this Investigation

5 A short time ago I released my report, *From Hope to Despair*. It dealt with a complaint that the Ministry of Health and Long-Term Care was refusing to fund a life-saving drug for a child with an inherited metabolic disorder. While conducting that investigation my investigators uncovered a deeply-troubling, even chilling, electronic message penned on July 20, 2005 in reaction to that investigation. That message:

> … pointed out the potential for this investigation [*From Hope to Despair*] to get into the whole IMD [Inherited Metabolic Diseases] program, including the screening issue, where … there have been 5 deaths from MCAD[2] and coroner opinions voiced.

6 It was that red flag, and what we uncovered in researching *From Hope to Despair*, that spurred this current investigation.

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[2] The actual acronym is MCADD, which stands for Medium-chain acyl-CoA dehydrogenase deficiency.

7 To be more precise about what was uncovered about the IMD Program and newborn screening while investigating *From Hope to Despair* I learned that the IMD Program, which was responsible for treatment protocols for inherited metabolic diseases and which had taken on responsibility for newborn screening issues, was moribund. A medical Advisory Committee to the IMD Program had ceased to operate in 2003. I learned that the program floundered not as the result of some sage reorganization plan, but because of bureaucratic inertia and turf-disputes, and I learned that the Committee's Chair resigned out of frustration that the Committee's expert advice was being ignored. I learned about the consequences of that failure, consequences that went far beyond the omission to make timely and appropriate decisions about drug-funding for children with inherited metabolic diseases. I discovered that children were being put at risk of severe disability and death because of Ontario's failure to undertake possible, affordable steps to detect inherited metabolic disorders through newborn screening. I learned, in particular, that MCADD, the condition referred to in the electronic message we had discovered, is an inherited metabolic disorder that, if detected early by newborn testing, can be treated by diet and other means and that if not diagnosed a child with MCADD will go into crisis and become severely disabled or die. This disturbing reality was made even more graphic and troubling when I discovered that while Ontario was not screening newborns for MCADD, it was screening sudden infant deaths for the condition and for other inherited metabolic disorders, and I learned results obtained by the Coroner disclose that, in Ontario, even leaving aside other testable conditions, five deaths a year are related to the failure to detect MCADD alone. Children were dying because Ontario could not or would not get its act together.

8 Given the critical nature of this issue I initiated an investigation on August 11, 2005 into the Ministry's apparent failure to properly administer newborn screening in Ontario. This investigation was conducted by my Special Ombudsman Response Team (SORT). A team of six personnel, including investigators and senior counsel, undertook the work. SORT investigators interviewed 17 families who have been affected by the loss of a child or had children who suffered severe disability as a result of disorders screened in newborns in other jurisdictions but not in Ontario. A number of interested individuals and associations were also contacted. Four senior officials from the Ministry of Health and Long-Term Care were interviewed, as well as a former Ministry official. Six medical specialists and other experts gave generously of their valuable time, providing my investigators with information about their experience with newborn testing. The Deputy Chief Coroner was also interviewed.

9 The team reviewed approximately 5,000 pages of documentation and interview transcripts.

10 All formal interviews were tape-recorded.

11 This Report, *A Right to be Impatient*, is the result of that investigation and it confirms that things are even worse than I had imagined. Estimates the Ministry obtained indicate that of the approximately 130,000 children born in Ontario each year, as many as 50 children annually become disabled or die because our newborn testing program has stagnated. This is a stunningly dismal, even shameful record, one that the Government simply has to improve upon and learn from.

History of Newborn Screening

Descending from the Summit

12 Over forty years ago Ontario was at the vanguard in newborn screening. In 1965 it was a world leader when it introduced universal testing for a condition commonly referred to as PKU. The medical name for this inherited metabolic disease is phenylketonuria. If PKU goes undetected it can result in severe developmental disease. The remarkable thing is that if diagnosed, PKU can be treated with diet alone. And the test for PKU is simple and unobtrusive. Blood, obtained by pricking a baby's heel will show the disease. Because of newborn screening hundreds of Ontarians have avoided at little expense the limitations and heartbreak of degenerative disability.

13 In 1968, flush with the success of the PKU testing program, an Advisory Committee of the Ministry of Health on Screening Inherited Diseases in Children was created by Order in Council. This Committee was composed of various medical specialists and representatives of the Public Health Branch of the Ministry. It looked at the time as though Ontario would stay abreast of new developments and remain as current as possible in its testing programs.

14 This Committee's work indeed led in 1978 to use of the pin prick blood sample to test newborns for another condition, congenital hypothyroidism, (CH), which if detected can be treated, again preventing severe disabilities. Newborn screening discloses between 37 and 40 cases of CH per year in this Province.

15 After reaching this pinnacle, things began to decline.

16 Initially, it was simply a case of ignoring advice from the Government's expert advisory body. In 1991 the Advisory Committee of the Ministry of Health on Screening Inherited Diseases in Children tried, without success, to add Congenital Adrenal Hyperplasia (CAH) to the list of newborn tests. This condition, an endocrine disorder, can lead to disability or death if undetected. The recommendation met with delay, and then, even after the Committee met in 1992 with Senior Ministry Officials on the matter, no action was taken.

17 Then, a year or so later that advisory body suggested that the Government of Ontario begin testing for Sickle Cell Disease (SCD). This is an inherited blood disorder which can cause tissue damage, stroke and leave sufferers very vulnerable to bacterial infections. Early diagnosis and prophylactic use of penicillin, particularly during a crucial window when the child is between two and three months of age, can be effective in treating SCD, reducing death or illness by 85 per cent.

18 After a couple of years of procrastination the Advisory Committee's recommendation led in 1995 to a 10-week study that revealed significant incidents of the disease or its traits. Of 7,554 specimens from the Greater Toronto Area, three cases of SCD and 80 cases of newborns carrying the trait of SCD were identified. A pilot project was recommended. After three years of inactivity, the call for a pilot project was renewed. Despite the results of the 10-week study and 10 years of effort by the Advisory Committee, the proposal for a pilot project has languished and nothing has been done.

19 Now a pilot study no longer seems necessary. The need for SCD screening has clearly increased with the demographics of Ontario, particularly in Metropolitan Toronto. The incidence of the disease is now 13.2 out of 100,000 live births, or 20 children a year. For West African children, the incidence is even higher, approximately one out of 100 live births. Yet SCD is still not tested for in newborns other than through the independent initiative of the Scarborough Hospital which conducts its own SCD screening at a cost of $2.50 to $3.00 per sample using high pressure liquid chromatography which is now available in many hospitals. Had the Advisory Committee recommendations been acted upon in 1992, well over 100 children and families could have been spared incredible anguish. In a truly ironic twist, the new Ministry of Health Promotion's website states:

> Early diagnosis of Sickle Cell Anemia in children is extremely important. A simple, inexpensive blood test, called *hemoglobin electrophoresis*, can be used on newborn infants to screen for the condition. This test is done at the same time and from the same blood samples as other routine newborn screening tests.

20 Perhaps the new Ministry of Health Promotion should share its wisdom directly with the Ministry of Health and Long Term Care. It strikes me that by posting this admonishment on its website it is preaching to a very converted public audience.

21 In September 2002 yet another condition became the focus of a futile call for testing. This time it was MCADD, a disease that has claimed a total of seven children from families that we interviewed during this investigation.

22 In Ontario, inertia and resistance had replaced initiative.

23 Ironically, the lost commitment to preventative medicine in the newborn testing area was occurring at a time when huge advances were being made in the ability to test newborns. Specifically, during the 1990's Tandem Mass Spectrometry (TMS) was developed. This technology allows for simultaneous multiple screening, enabling many disorders, including MCADD to be identified using a single blood sample.[3] By the late 1990's Tandem Mass Spectrometry had been shown in the U.S., the U.K. and elsewhere to be highly effective and cost beneficial in the identification of infants with a wide range of treatable inherited metabolic disorders.

24 In March 2001, the Advisory Committee recommended that Ontario adopt Tandem Mass Spectrometry for newborn screening. Based on its report, the Ontario Medical Advisory Secretariat was asked to review the issue.

25 A September 2002 report of the Ontario Medical Advisory Secretariat extols the benefits of Tandem Mass Spectrometry. The Report, *Neonatal Screening of Inborn Errors of Metabolism using Tandem Mass Spectrometer* recites how this technology enables 25 inborn errors of metabolism to be detected with precision, using the same dry blood spot. In terms of cost, while it would require enhanced infrastructure for result interpretation and reporting it was observed that this

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[3] CAH cannot be tested for using Tandem Mass Spectrometry. Other techniques are utilized. It is nonetheless possible to use Tandem Mass Spectrometry equipment to identify SCD, although SCD cannot be tested for simultaneously with inherited metabolic diseases, and Tandem Mass Spectrometry is not a common way to test for this condition.

would not add significantly to Ontario's testing program. In terms of benefits, early detection through screening and simple treatments could avoid mortality and prevent or reduce mental retardation. The Report noted that as of 1998 twenty-six states in the United States were using Tandem Mass Spectrometry for newborn screening. In Canada, by 2002, British Columbia, Saskatchewan and Nova Scotia were using the technology, while Prince Edward Island and the Yukon Territories had universal testing programs in place utilizing the technology of provincial neighbours.

26 To signal where things now are, three years later Ontario is still only testing for two conditions and does not use this available technology. More is the irony. In the 1990's a company in Concord Ontario, MDS SCIEX, began manufacturing and distributing Tandem Mass Spectrometers to other jurisdictions for the purpose of newborn screening. We in Ontario make the technology and sell it to others but do not use it ourselves. It is as if a phone company used a telegraph while marketing its phones.

27 It is not as though there is any controversy about the efficacy of the testing. Given the available technology, on March 8, 2005, the American College of Genetics identified 29 conditions for which newborn screening should be mandated in the United States. Its recommendations have been endorsed by numerous organizations, including the March of Dimes. By last spring, two states legally required the use of Tandem Mass Spectrometry screening, removing the decision from the exigencies of administrative discretion. By that time 29 states were testing for MCADD and 17 others had expanded their testing using the technology.

28 Tandem Mass Spectrometry is used around the world. It is employed for newborn screening in Germany, Italy, South Africa, Australia (New South Wales and South Australia), Japan, South Korea, Saudi Arabia and Costa Rica.

29 As for Canada, Saskatchewan is among the world leaders. It tests for 28 inherited metabolic diseases. Prince Edward Island and Nova Scotia test for 10 conditions, and are running a pilot project that includes another four. Manitoba tests universally for six conditions and conducts further "targeted" screening. British Columbia tests for four conditions while Alberta, Newfoundland and Labrador test universally for three. Quebec, which uses newborn screening to test for three conditions, has an additional program for urine-impregnated filter paper testing at 21 days of age for amino acids and organic acids that detects 14 disorders that need early medical intervention and nine more that need mostly counseling and surveillance. As for us in Ontario, pending the implementation in the next year of current initiatives, we are in last place, still testing as though it was 1978.

30 The descent from the pinnacle is not, however, just some simple contest producing only bragging rights to the winner and humility for the loser. It is a health care calamity. And we have known it for a long time.

Pleadings and Warnings

31 Documents obtained in our investigation show that over the last three years in particular there have been frequent calls made for increased newborn screening. What is striking about these attempts is that, in an effort to drive home the urgency of the situation, the perils of not testing, including health, legal and ethical risks have been featured.

32 The primary champion of this effort has been Dr. Joe Clarke, Head of Metabolic Genetics at the Hospital for Sick Children, who has agitated for increased testing both on his own behalf and, until 2003, in his capacity as Chair of the Ministry's Advisory Committee.[4]

33 In January 2002 Dr. Clarke wrote to then Minister of Health and Long-Term Care, Tony Clement, encouraging the expansion of newborn testing. He familiarized the Minister with the increasing use internationally of Tandem Mass Spectrometry and noted that he saw four to six infants and children every year who suffered long periods of illness, or experienced brain damage or even death as a result of the failure to detect metabolic diseases that are easily treatable. In April 2002 he wrote to the Minister again, repeating his earlier comments and adding that he understood that the Ministry of the Attorney General had retained a private American contractor to obtain postmortem tests results on children. By this time, the Coroner's Office was beginning to test for inherited medical conditions of children dying suddenly and unexpectedly under the age of two.

34 Then in September of that year, the Medical Advisory Secretariat issued its report, *Neonatal Screening of Inborn Errors of Metabolism using Tandem Mass Spectrometer*, which also recommended expanding newborn testing. One of the versions of this Report that we received estimates that in Ontario the failure to detect inborn errors of metabolism other than PKU and CH results in 20-25

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[4] In this latter capacity he had less clout than no doubt had been envisaged when that Committee was proclaimed. By 1994 the Order in Council that had created the Committee was allowed to expire, and the Committee was soldiering on unofficially without any formal mandate.

children per year becoming moderately mentally handicapped and 20-25 children dying of inborn errors of metabolism diseases.

35 By June 2003, other measures were being tried to cajole an evidently lethargic government. Dwight Duncan, a Liberal Member of Provincial Parliament introduced a Private Member's Public Bill that would require the Government to expand newborn testing using Tandem Mass Spectrometry. He was motivated not by a report or foreign development but by the tragedy of one of his Windsor constituents. She had been lobbying for expansion of newborn testing since she lost her 8-month-old granddaughter to MCADD in 2000. Since Ontario did not test newborns for MCADD it was not until the autopsy results were obtained in 2001 that the cause of death was known. The Bill did not progress beyond first reading.

36 By this time, Dr. Clarke had had enough. In October he resigned as Chair of the Advisory Committee, citing frustrations stemming from a lack of policy home for the Committee. While the Committee remained, without either official status or a Chair it was effectively rendered defunct.

37 By late Fall 2003, the newborn testing issue was being identified in some quarters not only as a health care issue, but as a looming government crisis. Confidential advice was prepared for the Minister in which the Medical Advisory Secretariat estimates of 25 disabilities and 25 deaths per year were repeated, and caution was given that legal action and media advocacy could be anticipated. Still nothing was done.

38 On January 12, 2004, Dr. Clarke, now in his private capacity, wrote to George Smitherman, the current Minister of Health and Long-Term Care. He repeated information that he had provided to the former Minister, again referencing that yearly he saw four to six infants and children who had suffered death or disability as a result of failure to detect inherited metabolic diseases that are easily treatable. He referred to the report of the Medical Advisory Secretariat and to the fact that its recommendation appeared to have become stalled within the Public Health Branch of the Ministry. Nothing seemed to happen. Dr. Clarke wrote the Minister again on May 10 and May 27, 2004, stating that it was a matter of some urgency and requesting that the Minister personally look into the matter of newborn testing.

39 Dr. Clarke's persistence may have inspired a response. On May 27, 2004 a meeting was held on Mass Tandem Spectrometry of key areas of the Ministry of Health and Long-Term Care (including the Hospital Branch Priority Programs, Integrated Programs and Policy, Laboratories Branch, Medical Advisory Secretariat and the Public Health Division). In documents prepared for the meeting the Medical Advisory Secretariat said there was no reason to delay moving forward with the introduction of Tandem Mass Spectrometry to await an anticipated review given its 2002 report, which was confirmed by a similar UK Report in March of 2004.

40 Then on June 2, 2004, a newborn screening workshop was held by another Ministry of Health and Long-Term Care Advisory Committee, this one on Genetics. The recommendation coming out of that workshop was that the current practice of newborn screening in Ontario should be reviewed and updated to meet the ethically and legally required standard of care as established in other provinces and comparable jurisdictions in the developed world. It was noted that failure to do this constituted a breach of ethics and placed the government at risk of legal liability for preventable morbidity and mortality in infants and children. On the same date Dr. Sandra Bennett, administrator of the Inherited Metabolic Diseases (IMD) Program, sent an electronic message to Dr. Cairns, the Deputy Chief Coroner, following up on a previous discussion about sudden infant death and MCADD.

41 Around the same time, the legislative route was being tried again. On June 15, 2004, John Baird, Conservative Member of Provincial Parliament, introduced a Private Member's Public Bill for first reading to expand newborn screening for disorders that can be detected through Tandem Mass Spectrometry. One of Mr. Baird's constituents, Tammy Clark, had lost her nine-month-old daughter, Jenna, suddenly and unexpectedly in November 2002. It was not until post-mortem testing had been conducted by the Coroner's Office that the family learned that Jenna had died as a result of MCADD.[5]

42 On June 17, 2004, Dr. Clarke wrote to the Minister of Health and Long-Term Care again, referencing Mr. Baird's Bill and urging him to examine the recommendations that would emerge from the workshop on newborn screening.

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[5] On June 13, 2005, Mr. Baird's Bill regarding newborn screening was referred to the Standing Committee on Regulations and Private Bills. It is anticipated that a public hearing will be held in late September 28, 2005 to consider the Bill.

43 In an October 12, 2004 Ministry briefing note to the Assistant Deputy Minister it was again noted that the failure to detect inherited metabolic diseases other than PKU and CH, results in 20-25 deaths annually.

44 It is therefore beyond any question that successive governments have known about the problem. Astoundingly, even shocking morbidity statistics, threats of lawsuits and the prospect of public lobbying failed to provoke discernible governmental reaction, save for the long-shot efforts of two opposition Private Members' Public Bills presented by opposition members of Provincial Parliament who, through the suffering of their constituents, had come face to face with the reality of not testing.

The Slow Climb Back

45 To its credit, the Government of Ontario has now resolved to change things. The infrastructure is being set up for Tandem Mass Spectrometry testing, and promises have been made for substantial increases in the number of conditions tested. What prompted the change? It was not simply information, as it is evident that successive Governments of Ontario have been armed for a long time with the case for increased testing, and with knowledge of the consequences of not moving forward, yet they failed to move. The most dramatic catalyst for change was that the reagent used by the Province to conduct the test it uses to detect PKU in newborns will no longer be available after the end of 2005. While the Province has dithered, science and knowledge moved forward apace to the point where Ontario's once-prized testing program has become obsolete. What gave the retirement of the reagent particular urgency is that the Ministry discovered that it only had enough stock of the reagent to maintain testing until the end of March 2006, and that the lead time for converting to Tandem Mass Spectrometry is estimated to be between 12 and 18 months.

46 Still, it would be an exaggeration to say that this intelligence galvanized the Government into immediate and decisive action. Initially the steps it took as it began its ascent from the bottom of the newborn testing world were sluggish to the point of being laboured. At a time calling for quick decision, things were put under advisement and deliberation, deliberation that would last for eight months and be conducted without organized, expert advice - on November 16 a representative of the remaining members of the Advisory Committee had written to the Ministry stating that the Committee had disbanded.

47 Without the assistance of that Committee, a November 2004 policy paper took a conservative tact, suggesting that consideration be given to using Tandem Mass Spectrometry, but only to test for the two usual conditions, PKU and CH, as well as new testing for MCADD until criteria for further testing could be developed. Then in December 2004 the Government sent a questionnaire out to hospitals and specialists seeking additional information about the application of Tandem Mass Spectrometry in newborn screening.

48 Meanwhile, the heat was being turned up. On February 14, 2005, Dr. Jim Cairns, Deputy Chief Coroner advised the Ministry that an inquest was being planned into newborn deaths due to MCADD. On April 11, 2005, he wrote to the Minister requesting an update on the Government’s plans for newborn screening. He advised that his office screens approximately five dead children a year with MCADD. On May 1, 2005, the Ministry responded, advising that it was examining the newborn testing program.

49 Then the Ontario Medical Association added to the call for movement. The President wrote to the Minister calling for a review of newborn testing, and he too was told that the program was being reviewed.

50 An internal Ministry memo of April 20, 2005 calls for quick action because of the threat of delay to the ability to conduct PKU testing. It expresses “concern on the timing issue as it looks like another task force will be struck, meeting held, etc. Not to say that this is not important but it is extremely important to have MSMS [Mass Spectrometry] up and running considering the supply of PKU reagents and the one year time line to bring MSMS on line.”

51 It was not until the end of May 2005, that the matter was referred to an internal expert body, the new Newborn Screening Subcommittee of the Ontario Advisory Committee on Genetics. Dr. Clarke was appointed its Chair. It was decided at that inaugural meeting of that Committee that MCADD should be added to the testing panel as soon as Tandem Mass Spectrometry was implemented in Ontario. It was agreed that a draft Request for Qualifications would be ready by the week of May 30, 2005.

52 Then political pressure was added to the call for action. On June 1, 2005, the Save Babies Through Screening Foundation of Canada (founded by Tammy Clark), along with the Sickle Cell Association and the Thalassemia Association, went public. They held a press conference at Queen’s Park to bring attention to the cause of newborn screening on the anniversary of the introduction of PKU screening in Ontario. By this time, parents of children with metabolic disorders had been engaged for some time in a letter writing campaign coordinated through Dr. Clarke to urge the government to expand newborn testing.

53 Still, it was not until August 2005, almost a year after the Government received word that it had no effective choice but to move to Tandem Mass Spectrometry, that the Ministry sent out letters of invitation to selected hospitals asking them to bid to operate the laboratory for newborn screening using the technology. Responses were due September 2, 2005.

54 There is reason to be concerned as a result of all of the delay that has occurred about whether Tandem Mass Spectrometry will be up and running in time to assume PKU testing before the Provincial supply of reagent disappears. While the Ministry position is that Tandem Mass Spectrometry will be up and running for PKU tests by March 2006, there is reason to be skeptical. Government estimates for acquiring and calibrating the machines suggest that it will take good luck to meet that date, and the Ministry is aggressively seeking to obtain additional reagent to ensure there is no disruption in PKU testing. Attempts to secure custom orders from manufacturers have apparently failed, and the possibility of sending samples to other labs for testing is being contemplated. Unless Tandem Mass Spectrometry is up and running in time, the delay may prove costly. In a July 16, 2005 internal memo a Ministry Official acknowledges, “Our approach on procuring supplies is that the need to obtain the testing materials outweighs the costs.” Worse, delay caused by having to send the samples away can imperil the health of PKU children. An other memo of April 21, 2005 cautions, “Any delay in this interval impacts on the start of treatment which may result in a negative medical outcome for the newborn (e.g., the onset of mental retardation, acute medical crisis, etc.).”

Increased Commitment

55 According to a Ministerial Briefing Note dated July 22, 2005, the Minister committed to making an announcement possibly early September 2005 about the newborn screening program. At that time, the Government was considering purchasing three Tandem Mass Spectrometers and adding five additional tests in sequential fashion.

56 On August 11, 2005, I commenced my investigation into the Ministry’s administration of newborn testing in Ontario. I noted that our province ranked as a third world country in terms of the extent of newborn screening it conducts. The next day the Minister told media representatives that there would be more provincial funds to catch rare diseases in newborns and that "a reasonably modest" announcement would soon be made on funding.

57 On September 7, 2005, on the eve of my investigators meeting with senior Ministry officials, the Minister issued a press release stating that its universal newborn screening program would be expanded not just to include the five additional tests which were being considered on July 22, 2005, but by adding 19 tests for inherited metabolic disorders, including MCADD. It was noted that this was the first expansion of the program in 27 years and that it was based on a preliminary recommendation of the Newborn Screening Subcommittee of the Ontario Advisory Committee on Genetics.

58 On September 9, 2005, Senior Ministry officials advised my investigators that the announcement was “the first wave.” The first additions to newborn screening focused on tests that could be done using Tandem Mass Spectrometry and a recommendation came from the Advisory Committee around August 24, 2005. They advised that the new Advisory Committee would be working with experts in blood and endocrine diseases to determine what additional screening should be done for those disorders. It is expected that the final recommendations of the Advisory Committee will be made in December 2005.

Cautious Credit where Cautious Credit is Due

59 I applaud the Government of Ontario for finally making this a priority and for harkening to the expert advice it is receiving. The Government is doing the right thing by expanding the program, and I strongly expect that once its Advisory Committee makes its final recommendation, the number of conditions that will be tested for will grow further still. The State of Mississippi, for example, tests for 57 disorders. Our first step decision to test for an additional 19 is a marked progress, but it will not get us across the finish-line. Even though instituting new testing programs comes with an initial price-tag that (absent federal funding which remains a possibility) will fall to the budget of the government of the day, this kind of preventative medicine will be cost-effective in the long run. The costs of acquiring Tandem Mass Spectrometers will be between $300,000 and $600,000 per unit, and there are ongoing operating and maintenance expenses as well as the treatment costs that successful screening requires. Still, there is every reason to believe that these costs will be more than offset by the savings on treatment and support that would otherwise be required for those who are needlessly harmed by not testing and acting. Ministry documents suggest that the costs of testing will not be prohibitive, particularly given the benefits of newborn testing. The State of Utah newborn screening website estimates that the cost of treating a child who survives an acute metabolic crisis, depending on the circumstances, is $80,000 – $1 million in the first two years, so the point is made. Successfully screening for very few children could, in the long run, pay the annual costs of the entire program.

60 But any long-term financial benefits that will be realized pale by comparison to the human dimension. It has to be recognized that however long in coming, the Government’s plan and its newfound commitment to make newborn testing as good as it can be will save lives and prevent or reduce suffering. The first wave Ontario plan of testing for 21 conditions is expected to identify a disorder in one out of every 2000 live births, or 65 cases a year out of the 130,000 live births in this Province per year. If the Province of Ontario moves in the direction suggested by the American College of Medical Genetics, which is endorsed by major American health organizations including the American Advisory Committee on Newborn Screening, testing for 29 core conditions and 25 supplementary target conditions can detect problems in one out of 800 live births. The Province of Ontario is heading in the right direction, and if it both follows through and pays sufficient attention to international developments and experience, it will have arrived.

61 Even with this air of optimism, it would nonetheless be remiss for us to forget the past entirely. We have to learn from the mistakes that were made, both to ensure that the current plan will be implemented with continued commitment and sustained priority, and so that this kind of thing does not happen again. It is in a spirit of constructive criticism and not blame-mongering that we need to analyze the sad legacy of Ontario’s newborn testing.

What Went Wrong?

62 How did Ontario end up administering, for more than a quarter of a century, a dormant newborn screening system? How could a province as sophisticated and wealthy as our own remain so inert, so impervious to scientific development that its range of tests would stagnate and its techniques and methods of newborn screening become obsolete? More to the point, how did we sit back and do nothing to avoid preventable deaths and disabilities?

63 The “usual suspects” for inaction, “budget” limitations and mismanagement, each contributed to the failure of the newborn screening system. To be clear, when I refer to “budget” limitations I am not referring to sage “cost-benefit” calculations, or the establishment of appropriate spending priorities. I am referring instead to what I will call “line item accounting.” I am referring to the failure to act even when an expense is required because of disagreement within an organization over whose line item in the budget it should be attributed to. It can be likened to the irrational paralysis of a couple who permit the rain from a leaking roof to destroy their furniture because they cannot agree on who should pay for roof repairs. "Line item accounting" contributed to the failure of newborn screening.

64 So too, did mismanagement. The "mismanagement" I speak of is intimately but not exclusively related to budget considerations. Inertia, even in the implementation of sound and urgent policy, was caused by the "it's not our job" syndrome, by a "lack of leadership," by an "abdication of responsibility," and by a general lack of courage to display "an appropriate sense of urgency."

"Line Item Accounting"

65 The Inherited Metabolic Diseases (IMD) Program has traditionally been administered through the Public Health Division. The primary responsibility of the IMD Program's Advisory Committee is approving funding for medications, supplements and special foods for children with inherited metabolic disorders. While it was not responsible for actually administering the newborn screening program (a job that is carried out by the Provincial Laboratory under the Laboratories Branch), the IMD Program was also involved, until recently, in policy issues relating to newborn screening.

66 Initially, the IMD Program was not expensive. Indeed, it required no base funding and was "cash managed" as a line item under the Public Health Division's Infectious Disease Outbreaks budget. From the mid-1990s, however, the cost of the Program rose from about $500,000 a year to its present cost of around $4 million. That increase was due to improved diagnosis of metabolic diseases, and the increased costs of newly developed and more effective treatments. In other words, costs increased along with improvements in health care.

67 In the meantime, Government policy of fiscal restraint and downsizing put pressure on ministry budgets. The first newborn screening casualties of fiscal pressure were the attempts by the Advisory Committee to increase the range of newborn tests that would be conducted. We found, consistent with fiscal restraint policies, that in 1992 when the Advisory Committee met with Senior Ministry officials to discuss its recommendation to begin to screen for CAH, concerns were expressed about the resources required to follow up on test results.

68 As fiscal restraint was pursued more aggressively within Government, progress was not only impeded but stultified. Dr. Lesbia Smith, a former Ministry Official who oversaw the IMD Program explained that she was under a lot of pressure not to sign off on expenses. As a result the Sickle Cell Disease screening recommendation foundered. First, the Committee recommendation was met with the classic bureaucratic filibuster – it was told to provide further information. Then its repeated proposals for at least a pilot study went unheeded, and it never happened.

69 The second casualty of the policy of fiscal restraint was even more profound than the loss of specific screening initiatives – that casualty was the primary policy apparatus for newborn testing – the IMD Program itself.

70 As indicated, in 1994, the Order in Council creating the IMD Program's Advisory Committee expired, denuding it of its official status and no doubt much of its credibility as a body for inspiring Government policy. I cannot be certain that the decision to allow its official status to lapse was budget driven but in the climate this seems likely. The Order in Council certainly was not permitted to lapse because the Advisory Committee's job was done or had been effectively taken up by some other agency. Other than inattention, the ability to control the budget by denuding the Advisory Committee of its formal status seems the only possible explanation.

71 Whether this is so or not, what is crystal clear is that the IMD Program had become something of an unwanted child – even an orphan program - because of "line accounting" concerns. Dr. Smith said of the IMD Program, "it didn't fit anywhere and nobody wanted it - it was just an expense." Ultimately Dr. Smith was directed by her superior to divest the Program from the Public Health Division.

72 To be clear, the attempt to move the IMD Program was not motivated by a desire to improve organizational structures. As Dr. Smith told us:

> My personal opinion is that it is a public health program and it's a public health program anywhere in the world, which is why I fought tooth and nail to keep it in public health and I very reluctantly had meetings with other departments to try and do what I was told to do, and that is don't leave that room without leaving that portfolio there, or else.

73 Although she did not say so overtly, the implication is obvious. Public Health Division wanted this increasingly expensive initiative to be someone else's line item. The IMD Program became a "hot potato" which bureaucrats tried desperately to fob off on other divisions. In the ensuing years the energy that was spent on IMD Program issues was directed not at implementing its medical recommendations, but at trying to move the program onto someone else's budget line. It was in this perverse way that budget was permitted to undermine newborn testing.

Mismanagement – "It's not our job"

74 Dr. Colin D'Cunha, former Chief Medical Officer of Health wrote to Dr. Clarke in May of 2001 explaining that Advisory Committee recommendations were not the responsibility of the Public Health Branch and that the Laboratories Branch should be responsible for newborn testing. An attempt was made to move the IMD Program to the Hospitals and Laboratories Branch but the Hospitals and Laboratories Branch took the position that its role was limited to testing. In the meantime, nothing was done to implement Advisory Committee recommendations.

75 Then a failed attempt was made in March 2003 to have the Director of the Drug Programs Branch take on the IMD Program.

76 In October 2003 it was the Ministry of Children and Youth Services that was said to be a "good fit" for the program. Six months later the Ministry of Children and Youth Services rejected the invitation.

77 In the spring of 2004, Public Health Branch staff renewed their efforts to find a home for the Program under the new direction of Dr. Sheela Basrur, Chief Medical Officer. Ministry officials began to discuss transferring the screening program from the Provincial Laboratory to a hospital administered through Acute Services.

78 Finally, in October 2004 a meeting of Assistant Deputy Ministers from various Branches was scheduled to discuss newborn screening. As a result, the Integrated Policy and Programs Division was identified as the genetics-related policy lead for the Inherited Metabolic Diseases (IMD) Program, while the Public Health Branch retained administration of the funding for drugs, supplements and specialty foods. Still, it was not until May of 2005 that the Integrated Policy and Programs Division managed to get a subcommittee on newborn testing up and running.

79 For three and a half years – crucial years for the newborn testing issue - the IMD Program was an unwanted child, an orphan program that was treated as no-one's job.

80 Without question, this lack of a real policy home for newborn screening is largely responsible for the failure of the newborn screening program. When the Medical Advisory Secretariat reported in September 2002 that newborn testing should be expanded, a significant organizational challenge was posed and, because of the lack of a policy home for the Program, that challenge could not easily be solved. Dr. Sandra Bennett, who had assumed Dr. Smith's oversight role of the IMD Program, was told that before increased testing could be accomplished there would have to be a coordinated policy home to bring together the various sections of Hospitals, Laboratories, Public Health and Drug Programs – "it wasn't something that could be done piecemeal."

81 As a result, energy was not expended on implementing testing, which the Medical Advisory Secretariat identified as essential to save lives, fulfill ethical responsibilities and avoid lawsuits. Instead, energy was spent circulating a flurry of correspondence relating to who should take responsibility for the IMD Program and its newborn screening issue. As Dr. Levin of the Medical Advisory Secretariat commented in an April 5, 2004 electronic message, the policy decision rested with Public Health and Laboratories and had it been up to the Secretariat, it would have been implemented in 2002.

Mismanagement – "The need for leadership"

82 In a July 9, 2004 electronic message, John Garcia, Director of the recently created Chronic Disease Prevention and Promotion Branch, put things succinctly when speaking of the failure of the Government to act on newborn screening recommendations - "Needs leadership and executive decision." He later commented the same day,

> While I have not been around PHD [Public Health Division] long, my view was that someone needs to decide and get on with it.
>
> I am not sure that there is a champion for this yet. Nevertheless, it is an issue that needs resolution and I would like to see this on the MMC table – as there are implications for multiple areas (hospitals, labs, public health and drug programs to name a few). …I hope that it is on MMC before it becomes a media or legal matter.

83 Without question, Mr. Garcia is right. The disconcerting game of ping-pong over which Division within the Government should orchestrate newborn screening left this crucially important health-care project without effective leadership that could have solved the problems that implementation posed. No-one in a position of effective responsibility took control over the matter.

84 The importance of leadership within Government to the newborn testing issue can also be seen in the comments made by Dr. Smith when speaking of the failed attempts to advance the Advisory Committee's recommendations to implement a pilot study for Sickle Cell Disease. She said, "I wasn't high enough. I mean they had no champion."

Mismanagement – "Abdication of Responsibility"

85 While medical experts advising the Ministry were dedicated champions for newborn screening, they could only use their positions to make recommendations, they depended on those within the Ministry to adopt and implement Committee advice. Meanwhile, there was no person or body within Government charged with the responsibility of moving Advisory Committee recommendations forward. By assigning an advisory committee responsibility for instigating policy decisions and then not supporting it, the Government had effectively delegated its own policy making responsibility to a powerless external Committee, creating the illusion that someone was looking after things. As a result, instead of receiving assistance Dr. Clarke and his Committee were met with underdeveloped fiscal objections, procrastination strategies like requests for further information, and the distinct and accurate impression that they were not wanted by the Division they belonged to.

86 Not surprisingly, the Advisory Committee, which had been operating without any official status since 1994, became increasingly frustrated with the Ministry's failure to act on its recommendations. In October 2003, Dr. Clarke chose to resign citing frustrations stemming from the lack of a "policy home" for the Committee. The Ministry has advised that after this point, the Advisory Committee had essentially ceased to exist.

87 On November 16, 2004 the *coup de grace* was delivered. A representative of the remaining members of the Advisory Committee wrote to the Ministry advising that the Committee was disbanding as it had no official standing with the Ministry and it was clear that there was no change on those issues the Committee deemed critical to the diagnosis and management of inborn errors of metabolism. It was noted:

> Despite our clear advice and best efforts there has been no progress on introducing extended newborn screening with an appropriate audit system. We have been frustrated by a lack of transparency and communication with your office….

88 On December 14, 2004, a number of the former Advisory Committee members wrote to Dr. Basrur, Chief Medical Officer of Health stating:

> We have been and continue to be disappointed at the refusal of the PHD (Public Health Division) to acknowledge that universal newborn screening for the people of Ontario is most definitely a public health issue and is seen as such by most other jurisdictions worldwide.

Mismanagement – "An appropriate sense of urgency."

89 One of the most astounding features of the failure of the newborn screening program is that despite persistent and credible harbingers of doom, no-one within Government showed an appropriate sense of urgency. Since 2002 there was nothing short of a flurry of missives and events surrounding newborn testing that should have galvanized the Government into swift and decisive action. The Chair of the Advisory Committee wrote time after time to Ministers, both in his official and personal capacity, giving testimony about witnessing preventable death and disability and communicating chilling statistics about the extent of suffering. The Medical Advisory Secretariat made formal recommendations, affirming that children were pointlessly dying and needlessly becoming disabled and noting that there are ethical obligations and potential legal implications of not moving. Internal memos cautioned against lawsuits and advocacy in the press with its attendant political fall-out. Private Members' Public Bills were presented in Provincial Parliament in the name of grieving families. Press conferences were held by affected families and advocates.

90 Together all of this should have been a call to arms. Observers of this episode can be forgiven for asking, "What does it take to inspire decisive and swift government action?" One would have thought that someone would have stepped forward, not to refer the matter to some committee for further study, but to act definitively and with an appropriate sense of urgency. It was obvious decisive action was needed.

91 On July 8, 2004, Dr. Levin sent an electronic message to various Ministry officials making this essential point this way:

> … If MAS is needed to provide any further evidence-based support for this we would be pleased to help but *this does seem to be an area in which a quick decision is needed, given the potential impact of this additional screen on decreasing avoidable deaths in the first year of life and decreasing the number of mentally challenged children.* (emphasis added)

92 And what was the consequence of politicians and bureaucrats failing to sense the urgency and to dig in and find out what a mess that things were and to remedy the situation peremptorily? When interviewed by my investigators, Dr. Clarke spoke passionately about newborn screening in Ontario. In response to a question as to whether there would have been a difference in his practice if the Government had extended newborn screening in 2003 or 2004, Dr. Clarke commented that:

> … there would be some children alive who are dead now. I think that there would be other children who currently are disabled who would not have been disabled.

93 Dr. Clarke advised that he personally knows that 25 percent of the patients he has seen over the last 12 years with MCADD were dead when the diagnosis was made and another 25 percent were irreversibly brain damaged. The remaining 50 percent were identified because they had an affected sibling who had died or become severely brain damaged.

Lessons to be Learned

94 As I say, there are important lessons to be learned from this sorry episode, lessons that are important both to the future direction of newborn screening and for government generally.

95 First, while we all want fiscal responsibility, decisions based on what I have called “line item accounting” have to be avoided. Government responsibilities to citizens are owed at large, not in isolation by divisions of government. It is contrary to those “at large” duties for essential jobs not to be attended to because individual government sectors believe they are being tasked with responsibilities others should be discharging. The right thing to do when there is a sense that this is happening is to discharge those responsibilities faithfully and with commitment, while trying to achieve reorganization. It is never appropriate to abdicate those responsibilities while waiting for reorganization to occur.

96 Second, and closely related to the first lesson is that, the "it's not our job" mentality has to be suppressed whenever it is encountered, particularly in health care matters. Failure to take charge of a problem within one's reach can mean death or pointless suffering.

97 Third, those who have responsibility have to take ownership or possession of issues that they are assigned. They must show leadership.

98 Fourth, while it is always prudent to consult experts and it is frequently sage to have advisory bodies, outside experts and advisory bodies can never be left unsupported within Government. There has to be an internal champion whose responsibility it is to advocate and push for the recommendations that are made. This is not to say that advice received must invariably be followed but it is to say that advice received must invariably be evaluated on the basis of the strongest case that can be made for it, and there must be people who can bring the matter forward to those who will get things done if resistance is met.

99 Fifth, there are times when highly placed government officials must be decisive, taking charge of urgent issues and solving them resolutely, even erring on the side of action rather than inaction. Think about the calculus in a case like this one. If a decisive move is made to test for a condition and it proves over time that this testing was unnecessary, the decision can be reversed and all that will have been lost is a tolerable amount of money. If no move is made to test for a condition, and children die or are disabled as a result, it is too late to recover and the costs both human and financial will be staggering. Prudence does not always equate with deliberation and slowness. Erring on the side of caution sometimes means going ahead without all of the formal approvals and processes being completed.

100 Sixth, and intimately related to each of the foregoing concerns, is the point that is implicit in John Adams' call for parental involvement on advisory bodies – that someone needs to be there to remind bureaucrats that there are human beings who are affected in real and dramatic ways by the decisions that get made or do not get made. Decision making, particularly in the health care field, must always have a human face.

101 While I am certainly not in any way opposed to parental involvement, I am not yet convinced that it is required in order for Ontario's governors and civil servants to achieve appropriate compassion and context. Admittedly, it is clear that in the case of newborn screening the issue was abstracted and bureaucratized by some key players; if that had not happened, the newborn screening program would not have failed the way it did. In spite of this there is surely enough humanity in all of us that if we only stop from time to time and think in human terms of the consequences of what we are doing, we will do the right thing.

An Example of Appropriate Reaction

102 What is now happening in the case of newborn screening appears to bear testimony to what can happen when appropriate practices are undertaken. When the Ministry found even a temporary policy home for the IMD Program in 2005 within the Integrated Policy and Programs Division under the direction and leadership of Carol Apparthurai, and then gave support to the newly formed Newborn Screening Subcommittee of the Ontario Advisory Committee on Genetics, things began to happen apace. On May 25, its first day, the Committee recommended that MCADD be added to the list of conditions to be tested. By 22 July 2005, the decision had been taken within Government to purchase three new Tandem Mass Spectrometers and to add five additional tests to the two currently being conducted. Within a couple of weeks the Advisory Committee was in a position to recommend that a total of 21 tests be conducted (amounting to 19 new tests in total) and the Government, now indisputably apprised at all levels of the medical consequences of not acting, took decisive action and committed to add those 19 tests instead of the five initially proposed. And this is only the "first wave." The Advisory Committee is currently examining for possible newborn screening conditions that cannot be tested by Tandem Mass Spectrometry, including inherited metabolic diseases like G6PD and Galactosemia; endocrine disorders such as CAH; and blood disorders like Thalassemia and the long-ignored Sickle Cell Disease. While parents who have witnessed years of inaction are naturally suspicious, especially those whose children's conditions did not find themselves on the list of 19, all indications are that the Government will pay close attention to the Advisory Committee recommendations and that the Advisory Committee will look far and wide for best practices and precedents. Finally, things appear to be progressing as they long should have.

The Conclusion is Yet to be Written

103 The Ministry has advised that it should have all the recommendations for additions to the newborn screening program by December 2005. Tandem Mass Spectrometry is scheduled to be implemented by March 15, 2006, in time to pick up PKU as supplies for traditional testing expire. However, there are still a number of unknowns. The screening centre has not been identified. Equipment must be purchased and set up. Training must take place. There are also other issues relating to establishing a secure infrastructure for a renewed newborn screening program, an information management system must be created (currently the Provincial Laboratories system for tracking PKU results is manual), result interpretation must be considered, as well as education and issues around consent, counseling and medical management. On December 17, 2004, Dr. Clarke advised the Ministry that it would take 12 to 18 months to get the program up and running.

104 In addition, the actual time frame required to calibrate the Tandem Mass Spectrometers is unclear. The Ministry has stated its understanding that it might take up to a year to begin testing for all 19 conditions in addition to PKU yet Saskatchewan's experience was that it took substantially less time to begin testing for 27 conditions through Tandem Mass Spectrometry.

105 Given this period of transition and the fact that some latitude ought to be given to the Ministry to address issues and devise its own approach, I will not be making conclusions about newborn testing decisions, nor will I be making any immediate recommendations arising out of this investigation. I plan to wait for six months to give the Ministry time to proceed with its plans and to formulate an appropriate response to the observations I have made. In order to satisfy myself that things are still moving briskly I am asking the Ministry to provide me with an update on its progress in 90 days. If after the expiration of six months, the Ministry has not proven that it has reasonably addressed issues relating to newborn screening in Ontario, I will decide whether to issue a further report with conclusions and recommendations, or whether to conduct a full public hearing and subpoena all necessary witnesses.

106 In taking its next steps to address newborn screening, I urge the Ministry to heed the lessons learned from the sad history of newborn screening in Ontario. It cannot simply rely on medical specialists, who give time away from their busy practices, to be the catalysts for government policy development for newborn screening. Government must take the lead in planning and implementing newborn screening expansion. This must be a priority, and it must have a proper program home. Creative thinking should be applied to ensure that the Government area assigned the role of coordinating the multi-faceted aspects of newborn screening is secure and senior enough to be able to withstand the bureaucratic inertia that has plagued the newborn screening program in the past. This might even involve looking beyond the confines of the Ministry itself to related Ministries such as the new Ministry of Health Promotion for novel solutions.

107 While I will stop short of making recommendations at this time, I will offer this counsel in the interim. Those involved in the process can avoid lapsing into inertia or inattention or the temptation to leave the work to others by simply recalling that as a result of inertia, inattention and abdication in the past, children have needlessly died and been rendered disabled. If this realization does not motivate this Government to stay the course, nothing I recommend will ever do so.

André Marin
Ombudsman

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