LES 11

O B S E R V A T O R Y
ENVIRONMENTAL SCIENCE AND TECHNOLOGY (EST)
Teacher's Guide A
Second Year of Secondary Cycle Two

A QUESTION OF ETHICS OR SCIENCE?

STUDENT LOG

PROCEDURE AND EVALUATION: SSC2 – SCIENCE

Observatory / Guide
11129-A

Name: _____ Group: _____ EST

The case study

PRESS RELEASE
For immediate release

DOWN SYNDROME
The ethics of prenatal screening:
The Commissaire à la santé et au bien-être releases a consultation report

Québec, January 26, 2009 – Today the Commissaire à la santé et au bien-être [Commissioner for health and welfare] released its report ***Consultation sur les enjeux éthiques du dépistage prénatal de la trisomie 21, ou syndrome de Down, au Québec – Des choix individuels qui nous interpellent collectivement*** [***Consultation on the ethics of prenatal screening for Down syndrome in Québec: Private choices that call for public debate***]. The report is the fruit of a year of consultations on the ethical issues of prenatal screening for Down syndrome. . . .

"In light of the conclusions we have drawn from our consultations, we have identified a series of essential measures to take. During prenatal care, all women should be offered a prenatal screening test for Down syndrome (trisomy 21) along with the information they need to make an independent and informed decision. If a screening test indicates a high risk of trisomy 21, an amniocentesis should be performed to confirm the risk. In the event of a positive diagnosis, parents must feel free to decide, in full knowledge of the facts, whether the mother will carry the pregnancy to term. Parents must also receive support regardless of their choice. The quality of the information provided to the parents and the way it is communicated to them are important. The resources we will allocate to transmitting this information will reflect the priority we are giving this initiative," said Commissioner Robert Salois. . . .

The consultations

. . . The consultations revealed the following findings:

Prenatal screening for Down syndrome is of great interest to the public. The relevance of the consultation lay in the major ethical issues this question raises. The importance of parents' free will with regard to procreation was reiterated throughout the consultations. . . . To respect this principle and help parents make an informed decision about whether to continue a pregnancy, attention must be paid to the type of information they receive, the way in which it is communicated and the resources that are allocated to the communication process. The option of taking the test and the relevant information must go hand in hand. Finally, support for individuals with intellectual disabilities and their families could be greatly improved. . . .

– 30 –

Source: Portal of the Government of Québec, Press release from the Commissaire à la santé et au bien-être[1] [online], January 26, 2009 (accessed February 5, 2009). *[Translation]*

1 The mission of the Commissaire à la santé et au bien-être is to make relevant contributions to public debate and government decisions in the interest of improving the health and welfare of Quebeckers.

Name: _____ Group: _____ EST

The case study (*continued*)

MEMO

Saint-Hyacinthe, February 17, 2009

TO: Communications officers
FROM: Chief Executive Officer, Polyvalent
SUBJECT: New mandate

Since the Commissaire à la santé et au bien-être recommended that the government offer prenatal screening for Down syndrome, staff at the Lerougeur hospital have been receiving many requests for information on genetic diseases. The commissioner's report stresses the importance of distributing information to help parents make informed decisions, so the hospital has mandated us to produce pamphlets on a number of genetic diseases.

The hospital has identified four diseases that are the subjects of most of the inquiries it receives:

- cystic fibrosis
- lactic acidosis
- phenylketonuria
- Tay-Sachs disease

Parents want information about these diseases and want to know the risk of their child's having them or being a carrier. The pamphlets will describe the symptoms of the disease, the life expectancy of people suffering from it, the prevalence of the disease, available treatments, and screening tests. It will explain the risk of having a child who either has the disease or is a carrier in the following three situations:

- when both parents are carriers of the gene for the disease
- when one parent is a carrier
- when one parent has the disease and the other is a carrier (when it is possible for a person with the disease to have children)

I am assigning your team the production of these pamphlets, which will contain texts of no more than 600 words. You will find information on these diseases in the enclosed information documents.

Thanking you in advance for your invaluable cooperation,

Yours truly,

Jasmine Triplehorn
Jasmine Triplehorn, CEO

In this context, you will play the role of a communications officer who must produce a pamphlet about one of the diseases identified by the hospital.

Name: _____ Group: _____ EST

Creating the context

The chosen disease: _____

I ask myself questions

1. What is a prenatal screening test?

2. What is the role of the Commissaire à la santé et au bien-être?

3. What does the expression *prevalence of the disease* mean?

4. What is amniocentesis?

5. What does the phrase “the parents’ free will with regard to procreation” mean?

Creating the context (*continued*)

6. What is Down syndrome, or trisomy 21?

7. How is information presented in a pamphlet?

8. Who are the main players in this situation?

9. What are the ethical and social issues related to genetic diseases?

Creating the context (*continued*)

10. Which of the questions that future parents might ask should guide you in your information gathering for this case study?

I must

11. Reformulate the goal of the case study.

I think

12. What information must be provided in the pamphlet to help parents make an informed decision?

Name: ______________________ Group: __________ **EST**

Creating the context (*continued*)

What I know and what I must find out

13. Write the information you already know and the information you need to find out.

What I know	What I must find out

Creating the context (*continued*)

I prepare my work

14. Where will you find the information you need for this case study?

15. Define the main steps of your case study in chronological order.

Reflection

	Yes	No
Do I fully understand what I have to do?	☐	☐

Name: _____ Group: _____ EST

Gathering information

I do research

1. What is heredity?

2. What is a gene?

3. What is an allele?

4. What is a character trait?

5. What information is provided by a genotype?

6. What is the difference between a genotype and a phenotype?

7. What type of grid is used to evaluate the risk that a child will have a genetic disease or carry the gene for it?

Gathering information (*continued*)

8. What information do you need to use this tool?

9. What is the difference between a homozygous individual and a heterozygous individual?

10. What is the difference between a dominant allele and a recessive allele?

11. Define *protein synthesis*.

12. What is crossbreeding?

13. What is the difference between crossbreeding and cloning?

14. Highlight the information you consider relevant in your information documents. Copy this information in question 15.

Gathering information (*continued*)

I apply my research results

15. What information should be communicated to parents? Summarize it below.

Name: _____ Group: _____ EST

Gathering information (*continued*)

Gathering information (*continued*)

I apply my research results

16. For each of the cases described in the CEO's memo, complete a grid to evaluate the parents' risk of having a child who has the genetic disease or who is a carrier of the gene for it.

Scenario 1: One parent is a carrier of the gene for the disease.

Gathering information *(continued)*

Scenario 2: Both parents are carriers of the gene for the disease.

Name: _____ Group: _____ EST

Gathering information *(continued)*

Scenario 3: One parent has the disease, and the other is a carrier of the gene for it.

Reflection

	Yes	No
Do I fully understand the concepts related to this case study?	☐	☐

Name: _____ Group: _____ EST

Completing the case study

I make suggestions

1. Prepare the plan for your pamphlet. Summarize your main ideas.

2. Make your pamphlet.

Reflection

	Yes	No
Have I considered other approaches?	☐	☐

Name: _____ Group: _____ EST

Validating the case study

I justify my approach

1. Does your pamphlet answer all the questions parents would probably ask? Explain your answer.

2. What are the advantages of the presentation format you have adopted in your pamphlet?

3. Cite the sources you used to find your information.

Name: _____ Group: _____ EST

Validating the case study *(continued)*

4. How could you confirm the reliability of the information in your pamphlet?

My reflections

Do you support the proposal to offer all women prenatal screening for Down syndrome, accompanied by essential information for making an independent and informed choice?

Name: _____ Group: _____ EST

My evaluation

Use the evaluation grid on the following page to evaluate yourself. Write A, B, C, D or E in the "Me" column of the chart below.

SSC2—Makes the most of his/her knowledge of science and technology				
Criteria*	**Observable indicators**	**Me**	**Teacher**	**Comments**
1	**Creating the context** Definition of the goal and formulation of the questions for gathering information		☐ With help	
2	**Gathering information** Relevance of the information and the risk assessment		☐ With help	
3	**Completing the case study** Production of the pamphlet		☐ With help	
4	**Validating the case study** Justification of the pamphlet content		☐ With help	

*** Evaluation criteria**

1 Formulation of appropriate questions
2 Appropriate use of scientific and technological concepts, laws, models and theories
3 Relevant explanations or solutions
4 Suitable justification of explanations, solutions, decisions or opinions

Evaluation grid

Name: _____ Group: _____

EST

SSC2 Makes the most of his/her knowledge of science and technology

Criteria*	Observable indicators	A	B	C	D	E
1	**Creating the context** Definition of the goal and formulation of the questions for gathering information	The goal of the case study is very clear, and the questions for gathering information are relevant.	The goal of the case study is clear, and the questions for gathering information are relevant.	The goal of the case study is not very clear, OR the questions for gathering information are not very relevant.	The goal of the case study is not very clear, AND the questions for gathering information are not very relevant.	The work must be done again.
2	**Gathering information** Relevance of the information and the risk assessment	All the information is relevant, AND the risks are correctly assessed.	Most of the information is relevant, AND the risks are correctly assessed.	The information is not very relevant, OR the risks are not very well assessed.	The information is not very relevant, AND the risks are not very well assessed.	The work must be done again.
3	**Completing the case study** Production of the pamphlet	The pamphlet is very clearly written and presented. All the information is correct.	The pamphlet is clearly written and presented. The information contains a few minor errors.	The pamphlet is not very well presented, the writing is good, and the information contains several errors.	The pamphlet is not very well presented, the writing is not good, and the information contains many errors.	The work must be done again.
4	**Validating the case study** Justification of the pamphlet content	The justification of the content is highly relevant, and the sources are cited correctly.	The justification of the content is relevant, and most of the sources are cited correctly.	The justification of the content is relevant, but the sources are not always cited correctly.	The justification of the content is not very relevant, AND the sources are not cited.	The work must be done again.

*** Evaluation criteria**

1 Formulation of appropriate questions

2 Appropriate use of scientific and technological concepts, laws, models and theories

3 Relevant explanations or solutions

4 Suitable justification of explanations, solutions, decisions or opinions

Information documents

The disease

What is cystic fibrosis?

Cystic fibrosis (CF) is the most common fatal genetic disease affecting young Canadians. CF is a multi-organ disease affecting primarily the lungs and the digestive system. In the lungs, CF causes severe breathing problems. A buildup of thick mucus makes it difficult to clear bacteria and leads to cycles of infection and inflammation, which damage the delicate lung tissues. They must follow a demanding daily routine of physical therapy to keep the lungs free of congestion and infection.

In the digestive tract, CF makes it extremely difficult to digest and absorb adequate nutrients from food. Thick mucus blocks the ducts of the pancreas, preventing enzymes from reaching the intestines to digest food. Therefore, persons with CF must consume a large number of artificial enzymes (on average 20 pills a day) with every meal and snack to help them absorb adequate nutrition from their food.

How many Canadians have cystic fibrosis?

It is estimated that one in every 3600 children born in Canada has CF.

At the present time, approximately 3500 children, adolescents and adults with cystic fibrosis attend specialized CF clinics. . . .

What causes cystic fibrosis?

People are born with cystic fibrosis; it is a genetic disorder.

Approximately one in every 25 Canadians carries a defective version of the gene responsible for CF. A carrier has only one copy of the gene responsible for CF. Carriers do not have cystic fibrosis and can never get the disease. In most cases, they are not even aware that they are carriers because they do not have cystic fibrosis or any of its symptoms. . . .

Is there a cure for cystic fibrosis?

As yet, there is no known cure for CF, but there is real hope.

Comprehensive treatment programs have dramatically extended the lives of persons with CF, and many are living into their 20s, 30s and beyond.

As of 2002, the median age of survival of Canadians with cystic fibrosis is 37 years of age. The median age of survival is the age beyond which half of the CF population can be expected to live. . . .

Information documents (*continued*)

How is CF treated?

Treatment programs are tailored to individual needs and depend upon the stage of the disease and which organs are affected. Treatments followed at home generally include:

- tapping or "clapping" the chest and the back vigorously (percussion) or PEP (positive expiratory pressure) mask therapy or other forms of chest physiotherapy to help loosen the mucus which clogs the lungs
- taking pancreatic enzymes with all meals, to aid digestion
- taking nutritional supplements and vitamins to promote good nutrition
- taking antibiotics in pill, intravenous (IV) or inhaled forms to ease congestion and protect against and fight lung infection
- exercise

How is CF diagnosed?

. . . Increasingly, genetic tests are being used in the diagnosis of the disease. Genetic tests are also used to diagnose CF prenatally.

Source: Canadian Cystic Fibrosis Foundation, "What is Cystic Fibrosis?" [Web page], January 3, 2008 (accessed July 10, 2009).

Information documents (*continued*)

A fatal hereditary disease

Degeneration of vital organs

CF affects many essential organs, especially the lungs and digestive system.

In healthy people, mucus in the body is fluid and helps keep the lungs and respiratory tract clear by aiding the elimination of germs and dust particles.

In people who have CF, this mucus is thick and sticky. It obstructs the bronchioles, causing breathing problems. Air is blocked in the bronchioles, and the lungs are blocked with mucus. Bacteria accumulate and multiply in the obstructed bronchioles, causing serious, recurring infections, which, in turn, lead to the deterioration of lung tissue. Pulmonary problems are the leading cause of death in people with cystic fibrosis.

The mucus also blocks the small ducts of the pancreas, an organ near the intestines, under the stomach. The pancreas secretes digestive enzymes that normally make their way to the small intestine, where they work to digest food. When the openings to the pancreas are blocked with mucus, these enzymes cannot reach their destination. Food leaving the intestines is only partially digested, so some of its nutritional value is lost.

The symptoms

- difficulty breathing
- constant cough which expels thick mucus
- excessive appetite, with weight loss
- bowel disturbances
- skin which tastes salty
- repeated or prolonged bouts of pneumonia
- failure to thrive

Source: Association québécoise de la fibrose kystique, “Qu’est-ce que la fibrose kystique?” [Web page], June 8, 2009 (accessed July 10, 2009). *[Translation]*

Information documents (*continued*)

What is Tay-Sachs?

Tay-Sachs is a hereditary genetic disease that results in an enzyme deficiency. When the enzyme hexosaminidase A is missing or in insufficient quantities in a child's blood, lipids accumulate in the nervous system, leading to severe neurological disorders.

The disease was identified in 1881 by an ophthalmologist called Dr. Tay, and in 1887, by a neurologist called Dr. Sachs, who linked it to a hereditary neurological disorder. At that time, the disease was referred to as "amaurotic familial idiocy" (infantile idiocy).

Treatment

There is no known treatment for Tay-Sachs at this time. Only medical support can be offered to children with the disease.

Diagnosis

It is easy to detect the disease in children with a DNA blood test. An eye exam can guide doctors for prediagnosis: they look for a "cherry-red spot" in the white of the child's eye. This spot is visible from between the 4th and 12th week after birth.

Three different forms of Tay-Sachs

Infantile Tay-Sachs (Type 1)

The disease appears between three and six months. It is irreversible. Affected children begin to show signs of weakness and loss of muscle tone (hypotonia). They lose their balance and can no longer support the weight of their head, so they tend to topple over. They can no longer hold themselves upright, sit up, crawl or roll over. They lose their manual dexterity and cannot handle objects, hold a utensil or eat on their own. At 12 to 15 months, swallowing becomes difficult. They choke on food or liquids with increasing frequency. They become very sensitive to noise and experience extreme nervousness. Spasms and convulsions appear along with a tendency to epileptic seizures.

At about 8 to 10 months, affected children tend to hold their legs in a "frog" position when lying down. Nothing can be done to correct this problem. . . . Their sight may weaken to the point of blindness by 18 to 22 months. . . .

Affected children usually die between the ages of two and five. There are rare cases of children living up to and beyond the age of eight, but this is exceptional.

Information documents (*continued*)

Juvenile Tay-Sachs (Type 2)

Another form of the disease, juvenile Tay-Sachs, affects children between the ages of four and six. The effects are different from those of Type 1. The children develop normally until the age of four to eight, and then a major regression takes place. They rapidly lose acquired skills, such as feeding themselves, talking, swallowing and eliminating. Their feet turn inward, making walking difficult. The same symptoms as in infantile Tay-Sachs usually appear: behavioural problems, loss of coordination and a deterioration of basic acquired functions such as language, balance and the ability to feed oneself. . . . The average life expectancy for children with this disease is 12 years. In Québec, there is presently one case of a 16-year-old child living with the disease.

Adult Tay-Sachs (Type 3)

This form of the disease is often confused with Friedreich's ataxia. It appears around the age of 10, with the same symptoms as the other forms of the disease. Despite these constraints, life expectancy is fairly high.

Population at risk

Ashkenazi Jews, from eastern Europe, have high rates of Tay-Sachs disease in comparison with the general world population. One person in 27 or 30, depending on the source, is a carrier of the gene for the disease. One child in 2250 in this population is born with Tay-Sachs, while the world average is one in 250 000.

With a rate comparable to that of the Ashkenazi Jews, French Canadians also have a higher than average incidence of the disease. Screening for Tay-Sachs in certain regions of Québec revealed that the number of carriers of the disease is one in 15 in the Bas-Saint-Laurent regions (approximately 6.2 percent of the population). These figures are well above known statistics for Jewish populations. People in the rest of Québec are also affected, with cases in the Estrie, Saguenay–Lac-St-Jean, Gaspésie and Rimouski regions. With today's mobile populations, no region is spared this terrible disease.

The Cajuns of Louisiana are another population at high risk. Children suffer from this disease all over Canada and throughout the world. . . .

How to prevent Tay-Sachs disease

. . . The best way to prevent the disease is prenatal screening. . . . An amniocentesis in the 12th week of pregnancy will show whether the child has the disease. . . .

Information documents (*continued*)

Available care

Talk to your doctor before using any of these suggestions.

As mentioned above, Tay-Sachs is incurable, but affected children can receive care to make them more comfortable. Some medications reduce, or even eliminate, the occurrence of seizures and convulsions. . . . Parents must watch very closely how their children respond to medications and should not hesitate to consult their doctors about reducing or increasing doses. . . .

Eating problems can be corrected with a **nasogastric tube or gastrostomy**. The two solutions are effective for dealing with malnourishment and choking. . . .

Feeding through a nasogastric tube often causes complications because of secretions that run down the throat. Solid or liquid deposits can accumulate in the lungs and cause aspiration pneumonia.

Source: Fondation Le monde de Charlotte Audrey-Anne et ses ami(e)s, Adaptation of the text “La maladie” [Web page], 2004 (accessed April 11, 2009). *[Translation]*

Information documents (*continued*)

What is lactic acidosis?

Lactic acidosis is caused by an enzyme deficiency. Cytochrome c oxidase (COX) is an enzyme that provides energy to the billions of cells that make up the human body. In this disease, the enzyme is reduced or absent in many organs (kidneys, muscles, brain, liver).

The disease takes its name from the first test used to detect it in children: a dose of lactic acid. Lactic acid is found in higher levels in affected children than in healthy children. It increases because the enzyme cytochrome c oxidase is absent. To understand why, we have to understand the role this enzyme plays in the body. We need it so that the billions of cells that make up our bodies can produce energy. Each cell contains a nucleus where our genetic code is stored, including a small organelle called the *mitochondria*. Cytochrome c oxidase is usually found in the mitochondria, where it is part of a chain of energy production. When it is absent, the cell cannot produce energy as it should, so the body receives a greater demand for energy than the cell can provide. . . .

Symptoms in children

Several symptoms of the enzyme deficiency can be observed in affected children. They are floppier than usual and learn to sit up and walk later. During an episode of infection, the body experiences an increased demand for energy. If cytochrome c oxidase is missing, the energy level of the body drops, accompanied by a blood imbalance that causes acidosis (acidic blood). If the imbalance is significant, the child may die.

Treatment

Children should receive certain basic care. We strongly advise parents to protect children from exposure to cigarette smoke and carbon monoxide from cars because these substances can interfere with cytochrome c oxidase function, which is already deficient in these children. We also recommend that they eat often, with snacks between meals to provide the food energy they need throughout the day. Large, rich and fatty meals should be avoided because digesting them takes a lot of energy. . . . These children need to live in a healthy environment, with a balanced diet of small meals throughout the day.

Prevention of infections is also possible. Appropriate vaccination is essential. . . . As for medication, a number of treatments are being tested or planned. . . .

Prevention

A screening test is available for people with a family history of lactic acidosis and for those whose partner is a carrier of the disease-causing gene. Screening tests are usually done in the context of family planning. Carriers are not affected by the disease, so this genetic characteristic becomes significant only when they want to have children (or when their adult children want to conceive). A genetic counsellor can help you decide whether it is worthwhile to take a screening test. . . .

Source: Association de l'acidose lactique du Saguenay–Lac-Saint-Jean, Adaptation of the text "La maladie" [Web page], 2009 (accessed April 14, 2009). *[Translation]*

Information documents (*continued*)

Lactic acidosis

Leigh syndrome French Canadian type (LSFC) is an autosomal recessive disorder leading to human cytochrome c oxidase (COX) deficiency caused by mutations in the LRPPRC (leucine-rich pentatricopeptide repeat cassette) gene. It is a disease of energy production and metabolism. Patients show developmental delay, hypotonia, mild facial dysmorphism, high mortality, due to episodes of severe metabolic acidosis, and coma (Merante et al., 1993). There is marked tissue specificity in the severity of the enzyme deficiency. The median life expectancy is about 5-6 years of age. The disease is rare worldwide, but extremely common in the Saguenay–Lac-St-Jean and Charlevoix regions of Québec, where the carrier rate is one in 22 and the disease affects one in 2000 live births.

Source: Laboratory in Genetics and Genomics Medicine of Inflammation, Montréal Heart Institute, hospital affiliated with the Université de Montréal, "Leigh's syndrome French Canadian type (LSFC)" [Web page] (accessed July 10, 2009).

Information documents (*continued*)

Phenylketonuria

Life without proteins

Marie Marcotte has phenylketonuria (PKU), a genetic disorder. PKU is caused by a defect in a specific protein—an enzyme called phenylalanine hydroxylase, which converts the amino acid phenylalanine into another amino acid called tyrosine. This conversion takes place through a chemical reaction in unaffected individuals.

In a person with PKU, however, there is no chemical reaction. Because of this, people with PKU have a buildup of phenylalanine in the blood and other body tissues. If untreated, this can cause severe problems, such as developmental delay, mental deficiency, seizures, autistic-like behaviour and a peculiar body odour.

The Guthrie test

The Guthrie test for PKU, developed in the 1960s, was the first genetic screening test. Today, it is a routine test performed in all hospitals in Canada. The blood phenylalanine level can be measured using a spot of dried blood. The timing of the test is important; it should be completed after the first day and before the seventh day of life. If done too soon, low levels in the newborn can be masked by the presence of maternal phenylalanine, yielding a false-positive result.

The phenylalanine hydroxylase gene is located on chromosome 12. When it is mutated, phenylalanine builds up in the body. There are 400 different mutations of the gene, which result in different degrees of PKU.

A special diet

PKU is treated by eliminating phenylalanine from the diet. Phenylalanine is present in all protein foods, such as meat, eggs and milk. Smaller amounts are also found in cereals, vegetables and fruits. As soon as Marie was born, she was put on a severe diet. As an infant, she was given a special "milk" formula with no phenylalanine. As she grew up, her parents had to control, weigh and measure all of her foods. Marie stopped the diet at age six, the recommended practice at the time. Today, doctors recommend that the diet be followed for life, as some individuals who have stopped the diet have encountered problems later in life.

Not every person with this genetic disorder has the same degree of enzyme deficiency. Marie, for example, has a milder form of PKU. Because her body can process some proteins, her diet is more liberal. The diet for the most severe form of PKU eliminates all of the very high protein foods, since all protein contains phenylalanine. A synthetic formula is used as a nutritional substitute for the eliminated foods. The diet is also supplemented with special low-protein foods, as well as weighed or measured amounts of fruits, vegetables and some grain products.

. . . The incidence of carriers in the general population is approximately 1 in 50 . . . The incidence of affected people in the general population [is] 1 in 10 000.

When PKU is treated at birth, normal early growth and development take place. At one time, children with the severe form of PKU were destined to become mentally disabled and to spend their lives in institutions. Today, children with the disease are growing up normally. They are attending college and becoming productive adults . . ..

Information documents (*continued*)

PKU and pregnancy

Marie has six children, and she had to plan her pregnancies carefully. A few months before conception, Marie had to go back to a strict diet. Returning to the diet is very important. Women with PKU who have high levels of phenylalanine in their blood have a very large probability of harming their unborn baby.

During her pregnancies, Marie had to weigh and measure all of her food. She also took a supplement in the form of an amino acid drink three times daily, which she qualifies as "horrible-tasting." She had her diet checked every week by a dietician, who made sure that she was getting the right amount of calories and fat to keep her unborn baby healthy. Marie often had to add canola oil to her diet, to make sure she was taking in enough fat. She also had to take blood samples every week, to have her level of phenylalanine checked. Ideally, it needed to be kept below 6 mg.

Source: Canadian Museum of Nature, Using Genomics, "Living with a genetic disorder" [Web page], October 10, 2008 (accessed July 10, 2009).

Phenylketonuria

The symptoms

Soon after birth, a phenylketonuric child must adhere to a protein-free or very low protein diet. Otherwise, the child will develop severe and irreversible neurological disorders, including retarded mental and motor development, behavioural disorders, spasms, epilepsy, etc. Early management involving a low-protein diet will permit normal development. If this diet is not maintained throughout adolescence or adulthood, the patient may suffer from problems of concentration, fatigue and nervousness shortly after ingesting proteins. . . .

A rare disorder, with many variants

Phenylketonuria may be the most common metabolic disorder, but it is still a very rare disease. In North America, only one case occurs in 12 000 births. As rare as it is, phenylketonuria takes different forms.

The most common variety of this order involves a deficiency of the enzyme phenylalanine hydroxylase. A total absence of this enzyme is described as the "classic" and most severe form of PKU. At three percent of normal enzymatic activity, the condition is classified as "atypical" PKU. At three to six percent of normal activity, the syndrome becomes "moderate" PKU. A small presence of phenylalanine enzyme in the latter two cases helps break down a small quantity of phenylalanine.

Another form of PKU does not involve a deficiency of phenylalanine hydroxylase, but rather of that enzyme's cofactor, tetrahydrobiopterine (BH4), a molecule that helps phenylalanine hydroxylase break down phenylalanine.

Source: Association québécoise des maladies métaboliques du Réseau, "Phenylketonuria" [Web page] (accessed July 10, 2009).