

Case Study: (Knoppers)(Tassé)

Since the 1960s, blood samples from newborns have been collected and analyzed in the context of universal screening programs. Recently, an enhanced neonatal screening program has been developed in order to detect a greater number of diseases, such as sickle cell anemia, a genetically inherited disorder. In addition to the usual symptoms of anemia, those who suffer from sickle cell anemia are prone to severe pain crises, vulnerability to infections and reduced life expectancy. Usually, these analyses do not require that all the blood collected from newborns be used. Thus, once the tests are completed, the leftover dried blood samples are stored as residual waste biological material.

Part A

Charlene and Luc recently welcomed their first son, Mike, a beautiful baby boy. A few weeks after their child's birth, Charlene and Luc receive a call from the newborn screening program. Given the results of the analysis of their son's newborn dried blood spot, they are asked to undergo additional tests for sickle cell anemia. Fortunately, these tests indicate that Mike is not suffering from the disease. However, the parents learn that their baby is a carrier of the disease. While their child is not suffering from sickle cell anemia, he may experience minor symptoms in particular situations (for example at high altitude and while diving) and may transmit the defective gene to his children. Charlene and Luc are shocked: they wonder what to do with this information. Before having a child, they did not know anything about the universal newborn screening program. They are surprised that they were not asked to consent prior to the blood sampling. They are also concerned about informing (or not) their child when he will be sufficiently mature.

Questions - Part A

- What principles are at stake in the decision to include (or exclude) a disease in a neonatal screening program?

- Do you think that Charlene and Luc should be asked for their consent for neonatal screening? or for storage?
- What principles underpin the decision to inform parents of the carrier status of their child?
- In the future, should Charlene and Luc tell their child that he is a carrier?

Part B

Since the blood samples stored following newborn screening are considered residual waste material, Dr Danks, a researcher, would like to use them to determine the prevalence of a genetic mutation associated with a greater risk of developing diabetes. His study also aims to identify families and communities where this mutation is present and to propose preventive measures targeted at the community level. As all newborns are included in newborn screening programs, the blood samples provide a unique representation of the entire population. According to Dr Danks, using residual blood samples is less expensive and involves no risk or inconvenience to the children and their parents. Dr Raoul, who leads the newborn screening program in Dr Danks' hospital, refuses to give Dr Danks access to the resource. He believes that the objective of the screening program is to provide clinical care, not research. He also believes that the parents would disagree with the use of the dried blood samples for research purposes.

Questions – Part B

- What are the risks associated with the use of these blood samples for research?
- What benefits can you expect from the proposed research?
- What principles are affected when blood samples collected for clinical purposes are used for research?
- Who should have the right to authorize the use of residual biological material for research purposes?

Part C

Like all parents of children in the State of X, Charlene and Luc are invited to participate in a longitudinal biobank study. The main objective of this paediatric biobank is to study the distribution of rare genetic diseases and identify related factors (nutritional, genetic, social, economic, etc.) for better health system planning and prevention. Participation in this biobank involves the collection of information (via questionnaires on the child's health) and biological material (blood and urine). Parents will be re-contacted twice a year by the study staff in order to answer follow-up questionnaires so as to keep the biobank up to date. Charlene immediately agrees, but Luc refuses. He thinks it might be used against his child one day (e.g. insurance/employment).

Questions – Part C

- Who should consent to such research?
- What happens when the dissent of the other parent is known?

Part D

The longitudinal biobank study has been underway for fifteen years, and children who were recruited as infants are now teenagers. As these children mature, Dr Ronaldo, a researcher involved in the project, is uncomfortable about asking parents to answer the follow-up questionnaires since researchers have no way of knowing if the adolescents are aware of the project, or if they agree to participate. Therefore, Dr Ronaldo thinks that teenagers should be contacted and asked to consent (by themselves) to participate in this study. On the other hand, some members of the research team believe that requiring the consent of teenagers could jeopardize the success of the longitudinal study.

Questions – Part D

- What principles are involved in proposing to seek the agreement of teenagers? Or not to seek the agreement of teenagers?

- At what age do teenagers have sufficient maturity to understand a research project?
- What should be the role of parents in the decision to continue (or not) in the study?
- If the teenager refuses to participate in the study, what should the researcher do with his/her data and samples?

Part E

Dr Lafleur, a French researcher working on sickle cell anemia, would like to have access to these samples and data for his research.

Questions – Part E

- Can samples and data be transferred from one country to another?
- What are the benefits of using the samples and data from another country?
What are the drawbacks?
- Under which conditions can a researcher access/use the samples and data from another country?

Part F

Mike, now an adult, has been injured in a serious motorcycle accident, during which he suffered severe head trauma and multiple fractures. Due to partial paralysis and memory impairment, he has been hospitalized in a rehabilitation facility for over a year. Today, he is able to walk alone (using a cane), but still is burdened with short term memory problems. He is unable to remember recent events and is therefore unable to work. Since Mike cannot drive a car, Charlene helps him with different tasks, such as grocery shopping. Earlier last week, Mike received a phone call inviting him to participate in follow-up site visits for the longitudinal study in which he has been participating since birth. Mike immediately agrees to participate. However, Charlene believes that Mike is unable to consent and to meet all the requirements of the study. In addition, Charlene refuses to bring Mike to the hospital for the site visits.

Questions – Part F

- Is Mike capable to consent to participation in this study?
- Where is the border between capacity and incapacity to consent to research?
- Who can consent? Mike or Charlene?
- What are the difficulties in assessing capacity to consent in a longitudinal study?