Canadian College of Medical Geneticists
Guidelines for DNA Banking

Purpose of the Present Statement:
The purpose of the present statement is to recommend guidelines for DNA banking practices in clinical molecular genetics laboratories accredited by the Canadian College of Medical Geneticists (CCMG). It is expected to serve as a resource for molecular genetics laboratories as they produce their own individual policies for DNA banking. The statement was produced by the CCMG Molecular Genetics Committee in 2006-7 and approved by the CCMG Board of Directors on January 23, 2008.

Scope of Present Statement:
The present statement addresses guidelines for DNA banking in the context of medical genetic diagnosis in CCMG-accredited molecular genetics laboratories. The statement addresses both long-term banking of samples and storage of residual clinical samples remaining after diagnostic testing. This statement also applies to storage of RNA, blood or other tissues sent to molecular genetics laboratories for the expressed intent of extraction of genetic material for molecular diagnostic purposes. For simplicity, the use of the term ‘DNA banking’ throughout the document includes the storage of the aforementioned materials.

The present statement replaces the previous CCMG statement Policy Statement Concerning DNA Banking and Molecular Genetic Diagnosis\(^1\). Issues concerning the analysis of DNA samples are now part of the document CCMG Molecular Genetics Guidelines\(^2\). Details about consent for research are not covered in the present statement. The process used for obtaining consent for research purposes should follow the ACMG Statement on Storage and Use of Genetic Materials\(^3\) until a statement on this process is developed by the CCMG.

Points for Consideration:

1. Storage of long-term banked DNA samples versus storage of residual clinical DNA samples.

Banked DNA samples can be divided into two types:

1) Long-term banked samples, where the purpose of banking is extended storage of genetic material for potential future use in clinical molecular diagnostic testing.

2) Residual clinical samples, where genetic material remains after a current molecular genetic test is completed.

Long-term banking is typically performed so that samples can be retrieved for ongoing complex analyses, for future investigations in cases where initial tests may have been inconclusive, or future testing of other family members. Storage of residual clinical samples may occur for similar reasons, however, when samples are submitted primarily for immediate testing, it is not always made explicit to the depositor that samples may or may not be stored after such testing is completed. In addition residual clinical samples might be
used by laboratories for other purposes such as test development, validation or quality control.

2. **Banking policies, particularly with regards to storage of residual clinical samples, are often not adequately disseminated to referring health care providers.**

Although recommendations regarding long-term banking of samples were outlined in a previous CCMG guideline, the storage of residual clinical samples was not specifically addressed. Tacit assumptions by depositors have ranged from the expectation that residual clinical samples will be stored indefinitely to the assumption that they will be discarded as soon as the test in question is completed.

3. **Storage facilities are affected by the exponential growth in molecular testing.**

The previous CCMG guidelines recommended that samples be stored for 100 years, in duplicate in different locations. Compliance with these recommendations has become impractical due to the escalating volume of samples submitted to clinical laboratories.

4. **Analytical methods are more direct and sensitive than in past, leading to reduced need for future analyses on stored DNA samples.**

Direct mutation testing has reduced the need for long term storage of DNA samples, which may be unnecessary in many current molecular testing situations.
Recommendations for Laboratory Guidelines:

Laboratory directors have a responsibility to define and disseminate their policies for DNA banking, with respect to storage both of samples for long-term banking and residual clinical samples.

Referring health care providers have a responsibility to take into account laboratory banking policies, and to request appropriate long-term banking of samples when warranted. In the absence of special directives from the referring health care provider, the laboratory will be at liberty to invoke the communicated laboratory default policy with respect to disposal of residual clinical samples.

It is recommended that laboratory banking policies include information regarding:

A. Information Required for Informed Consent

The consent process is the responsibility of the referring health care provider. As such a detailed discussion of the consent process is not within the scope of this document. However, the health care provider requires information about laboratory banking policies in order to adequately inform the patient. For this reason it is recommended that the laboratory policies on DNA banking include information on the following:

1. Duration of sample storage, for long-term banked samples and for residual clinical samples.
2. Procedures to be undertaken for disposal of long-term banked samples at the request of a patient.
3. Purpose for which the samples may be used other than those requested by the depositor (e.g. anonymously for test development, validation or quality control).
4. Risks, such as the possibility that the sample may be lost or otherwise compromised, and the laboratory procedures to deal with such an outcome.
5. Who has access to the sample, and if samples are to be stored, who will have access in future (e.g. patient, next of kin).

As the consent process is the responsibility of the referring health care provider, when a laboratory receives a sample, it may assume that the referring health care provider has obtained consent, and further confirmation and documentation of such consent is not needed. Laboratory directors may wish to include on laboratory requisitions a general statement as a reminder that in signing the form the clinician confirms that consent has been obtained.

B. Deposition of Samples

1. The laboratory should accept samples only from health care professionals, and only when accompanied by proper documentation. Laboratories should develop policies outlining the minimal required information accompanying a sample and procedures
undertaken in the event that inadequate information is provided. Similarly, policies should be developed outlining the minimal requirement for labeling samples and the procedures undertaken in the event that a sample is improperly labeled.

2. Special consideration may be needed for samples received from remote or atypical sources or from other laboratories.

C. Storage of Samples

1. Duration of storage: Decisions regarding length of sample storage should be based on the initial reason for banking, and anticipated future use. Length of storage for long-term banking should be at least two generations (50 years\textsuperscript{4,5}). Length of storage for residual clinical samples is at the discretion of the laboratory, and may range from a minimal but defined interval after reporting of current clinical testing to indefinite storage. Laboratories should establish and clearly disseminate their respective policies to referring health care providers. A suggested default policy is storage of residual clinical samples for one year after the laboratory has reported test results.

2. Methods of storage: The relative stability of DNA and sensitivity of many assays allows a variety of options for sample storage to be considered, each with associated costs, limitations and risks. It is recommended that each laboratory determine their appropriate method of short and long term storage, with the overriding principle of ensuring suitable material for future testing.

3. Replication of storage: In the case of samples submitted for long-term banking, storage of the sample in duplicate, in separate locations is appropriate to ensure long-term availability of the sample.

4. Sample labelling: Coded labelling of samples is recommended. Each storage container should be labelled with a unique identifier, and all other relevant information tied to the unique identifier through data records.

D. Retrieval of Samples

1. Procedures for retrieval of samples from storage and associated handling should be developed.

2. Policies for destruction of samples at the end of the storage period should be developed. When a limited storage time has been defined, samples should be handled accordingly at the end of the storage period.

3. Policies concerning use of residual clinical samples for other clinical laboratory purposes should be developed. This may include the use of samples for test development, validation and positive controls in the clinical laboratory. Appropriate consideration should be given to removal of identifiers from samples before use in
these activities. Use of samples for research must be conducted under institutional Research Ethics Board oversight.

4. Sharing of samples with other laboratories: The need to provide samples to other laboratories is common, due to management of extended family members elsewhere or testing undertaken in another laboratory. Policies should address who may have access to stored samples (typically restricted to the patient depositing the sample or the guardian/next of kin of a deceased patient). It is recommended that whenever possible, and particularly for long-term banked samples, the laboratory responsible for shipping a sample should retain a back-up aliquot in case of sample loss, or for other future needs.

E. Information Linked to Samples

1. Policies should define how patient confidentiality will be maintained for banked samples. Information from the samples should only be provided to persons who have the right to receive the information under health information privacy legislation. Laboratory facilities maintaining banked samples and banking records must be appropriately secured from unauthorized access.

2. Laboratory policies should include actions to be taken in case of loss or compromise of banked samples. For long-term banked samples this may involve a letter to the referring health care provider indicating that the banked sample is irretrievable, so as to provide the opportunity to collect a replacement sample. Laboratory policies should also take into account actions to be taken in the case of an extended lapse of time between when the sample was submitted and the discovery its loss or compromise. Residual clinical samples may not require this level of oversight.

3. In general, laboratories banking samples are not expected to communicate directly with patients regarding new information or available testing, but via the referring health care professional. The laboratory should have a policy to define when and how it will communicate new developments to referring health care providers.
References


