



JS & RDF BIOBANK

As many of you already know, the Joubert Syndrome & Related Disorders Foundation (JS & RDF) is one of the founding members of the BioBank established by the Genetic Alliance. The GA BioBank will allow the JS & RDF to manage the collection, storage and distribution of biological samples and information needed by researchers.

The development of such a central repository of this “raw material” for research has been one of the JS & RDF’s highest priorities, and we are thrilled that this is now possible.

The JS & RDF made a commitment to the BioBank because the “old system”, where individual researchers each developed their own collections of samples, was not working. It was confusing and often burdensome for families, and it was not the most effective way to advance research. The Genetic Alliance founded the BioBank because many other Foundations similar to ours were dealing with the same frustrating roadblocks to progress. The JS & RDF Board spent countless hours agonizing over the pros and cons of the BioBank before making the major policy and financial decision to participate in the BioBank. We are all unreservedly in favor of this project.

We already gave some detailed information about the BioBank in the Rainbow newsletter. As a reminder, this information is repeated here, with some minor changes.

What is the BioBank and why is it worthwhile for the JS & RDF to participate?

The BioBank is a cooperative resource developed by the Genetic Alliance. The purpose of the BioBank is to help family foundations collect, store and distribute biological samples and clinical information that can be used for research into rare genetic conditions, such as Joubert syndrome. In the past, this work has usually been under the control of individual scientists.

The BioBank will solve some of the problems that have hindered research into Joubert syndrome in the past. Some of these obstacles have been:

- 1) When individual scientists spend time and money to collect samples (i.e. blood, DNA, other tissues) and information about a condition such as Joubert syndrome, they are often unwilling to share this material freely with other scientists. Science is competitive. This is often a good thing, because it inspires researchers to work harder. It is not in our best interests, however, for individual laboratories to “hoard” samples and information about a condition as rare as Joubert syndrome. If the JS & RDF collects and controls this material, we can distribute it freely to any scientist who has a worthwhile project. This means that families do not have to provide multiple samples for different laboratories.
- 2) If the JS & RDF spends the time and money to collect this material, and makes it available to scientists, this will encourage new investigators to work on Joubert syndrome. Right now, a scientist might be discouraged from starting a new project on Joubert syndrome because of the difficulty in collecting samples. This is especially true of young scientists who are just starting out, and these are precisely the kinds of energetic researchers we would like to enlist at the beginning of their careers.
- 3) The JS & RDF BioBank materials can be collected with a high level of quality and standardization. When individual laboratories collect material there may be inconsistencies that make it hard for samples and information to be shared (when laboratories are able to agree to share). The JS & RDF collection will also ensure that the samples are not lost or put into limbo if a laboratory loses funding or if a scientist

retires or stops working on Joubert syndrome.

4) In the past there has been confusion on the part of JS & RDF families as to which laboratories were collecting and/or keeping their samples. There have been inconsistencies in the quality of the consent process used in obtaining the samples. The JS & RDF BioBank will make it much easier for families to understand their rights as research participants and to know who has control of their samples.

How will the BioBank work?

Families affected by Joubert syndrome will be offered the opportunity to contribute to the BioBank. Participation is in no way mandatory for JS & RDF members, and those who do participate will be able to withdraw at any time. Participation in the BioBank does not mean that you cannot give samples to individual investigators or other research projects, but we think this is not in everyone's best interests. The fact that you have given samples previously does not mean that you should not contribute to the BioBank, because we want the BioBank to be the largest, most useful resource available.

Families participating in the BioBank will be asked to contribute a blood sample and a swab of cheek cells. These samples will be used to make DNA, which can be stored indefinitely. Blood may also be used to make cell cultures. These cell cultures can be used to produce more DNA, and they can be used to study biological processes. We may want to include other kinds of material in the BioBank, such as biopsy material, buccal swabs (cells scraped from the inside of the mouth), urine samples, etc. For now, the BioBank is only collecting material for DNA. We need samples affected and unaffected family members.

Families participating in the BioBank will also be asked to provide clinical information about their affected children. There may be a lengthy questionnaire requested, and we may try to gather certain medical records. We realize that many families have provided this material repeatedly in the past and we will make every effort to gather previously collected information. It is important to realize that this information is highly prized as a research tool, and the biological samples are of little value without associated clinical information. Establishing a comprehensive, widely shared collection of this information has the potential to be of great use to families, because it allows a better description of Joubert syndrome so that we can predict and treat problems more effectively.

The BioBank is not yet ready to begin collecting, organizing and storing clinical information. The JSF & R CD is currently supporting a project by Dr. Bernie Maria, who many of you know well. Dr. Marie has established a Joubert Syndrome Registry, which will gather and analyze detailed clinical information about individuals with Joubert syndrome. We are fortunate to have someone with Dr. Marie's expertise to develop such a resource. This "Registry" is distinct from the BioBank, but we will be able to coordinate the information in the Registry with the BioBank samples at least until such time as the BioBank has its own mechanism for collecting clinical information. Dr. Marie is distributing surveys and we hope you will participate.

What is involved if my family wants to contribute to the BioBank?

Samples for the BioBank can be collected by mail, using kits that will be mailed to families. We are offering the opportunity for families to contribute samples at this conference. As we said earlier, we do not believe it is in the best interests of the JS & RDF or member families to allow individual investigators to collect samples at the conference anymore, because there have been conflicts over how these samples will be shared.

Anyone contributing a sample for the BioBank will be asked to sign a consent form that spells out the purpose of the BioBank, the pros and cons of participating, and the rules by which the BioBank is operating. We strongly encourage everyone to read the consent forms carefully and to ask questions about any of their concerns. Of course, this is true of any consent form you are asked to sign by anyone soliciting your participation in research.

Samples and information collected by the BioBank will be "anonymized", which means that all identifying

information will be removed and a coded number will be used for identification. The JS & RDF will retain the codes necessary to link samples to individuals, but this information will not be provided to investigators requesting material from the BioBank. This will protect participant's privacy. It will also make it easier for investigators to gain access to the material because they will not have to address privacy issues when they submit their research procedures to their own institutions' Human Subjects Committee.

How will samples from the BioBank be made available for research?

The JS & RDF will appoint an oversight board to make decisions about distribution of material from the BioBank. This board will have representatives from JSF members and from the scientific community. We will need to find scientists who are not conducting research on Joubert syndrome to serve on this board, so there will be no conflicts of interest.

Investigators who request material from the BioBank will have to meet the following criteria:

- 1) The project must address an issue of importance to members of the JS & RDF.
- 2) The project must have a reasonable likelihood of success.
- 3) The project must have an approved protocol from the investigator's Human Subjects Committee.
- 4) The investigator must agree to provide progress reports about the research, including negative results (things that didn't work) as well as positive results. The JS & RDF will respect reasonable requests to keep results confidential over the short term if their premature release would damage a laboratory's competitive situation.
- 5) Preference will be given to investigators who show a willingness to provide assistance to the JS & RDF in terms of participation in our annual conference, answering questions from JS & RDF families, and publication of written material about Joubert syndrome.

What should I do about participating in the BioBank if I have already given samples and information to other investigators?

As we have already said, participation in the BioBank is completely voluntary. However, we are very comfortable saying that it is in everyone's best interests for as many JS & RDF members as possible to support this project. The BioBank will be a major stimulant for advancing research about Joubert syndrome, both by supporting current investigators and by encouraging new scientists to enter the field.

Most JS & RDF members have already provided material to one or more investigators. This material is already being used for worthwhile projects and there is no reason to feel that these contributions were wasted. However, the value of these collections has been, and continues to be, diminished by difficulties in establishing cooperative agreements between laboratories. It is also a problem that the collections are fragmented and incomplete. In some cases, samples have been lost or are almost used up. Although it may be a burden, we are asking all JS & RDF members to consider contributing new samples to the BioBank. It is never fun to get a blood sample from any child, even without the added challenges posed by Joubert syndrome. We would not ask for this effort if we did not think it was important.

It is very important to remember that there are many rules protecting your rights when you participate in research. The JS & RDF BioBank is deeply committed to making sure that these rules followed rigorously. In particular, we want to remind you that your participation should always be voluntary. You should never feel as though you will be denied medical care or access to experts if you refuse to participate. You should always know exactly what your participation involves and you should have every opportunity to ask questions about the project and its procedures. It is unethical for any investigator to discourage you from participating in another investigator's legitimate research, or to insist that you contribute samples only to their laboratory. We encourage you to report any behavior that violates these standards to the board of the JSF & JSF and to the Human Subjects Committee of the offending investigator.

If you still have some questions, please feel free to contact:
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