1.0 **Purpose**
 This policy describes how investigators at the University of South Florida (USF) and USF Affiliates will submit data and materials as well as retrieve information from the Genome-Wide Association Studies (GWAS) repository database which is maintained by the National Institutes for Health (NIH). This policy also outlines the special considerations of the USF Institutional Review Board (IRB) when reviewing applications for the submission to or retrieval of data from the NIH database.

2.0 **Persons Affected:**
2.1 Principal Investigators
2.2 Research Staff
2.3 IRB Members
2.4 IRB Chairpersons or designees
2.5 IRB Staff and Administrators

3.0 **Policy:**
Under the NIH Policy on Sharing of Data Obtained in NIH Supported or Conducted Genome Wide Association Studies, institutions are responsible for certifying plans for the submission of genotype and phenotype data. It is the policy of the USF IRB that research involving GWAS be submitted to the USF IRB for the review of (1) plans for data submission to the GWAS data repository (dbGAP) and (2) the adequacy of the informed consent process and documents through which data were obtained should such data be submitted to the GWAS data repository.

4.0 **Definitions:**
4.1 **Coded:** Means that any identifying information (such as name or social security number) that would enable the investigator to readily ascertain the identity of the individual to whom the private information or specimens pertain has been replaced with a number, letter, symbol, or combination thereof (e.g., the code); and a key to decipher the code exists, enabling linkage of the identifying information to the private information on specimens.

4.2 **De-identified:** For purpose of this document, means that the identities of data subjects cannot be readily ascertained or otherwise associated with the data by repository staff or secondary data users (45 CFR 46.102(f)), the 18 identifiers enumerated at section 164.514(b)(2) of the HIPAA Privacy Rule are removed and the submitting institution has no actual knowledge that the remaining information could be used alone or in combination with other information to identify the subject of the data.

4.3 **Genome Wide Association Studies (GWAS):** The study of genetic variation across the entire genome that is designed to associate genetic variations (SNPs) with traits or with the presence of absence of disease of condition. Whole genome information, when
combined with clinical and other phenotype data, offers the potential for increased understanding of basic biological processes affecting human health, improvement in the prediction of disease and patient care, and ultimately the realization of the promise of personalized medicine. Competing GWAS applications must include a GWAS data sharing plan as part of the research plan (grant application) or outline why such data sharing is not appropriate.

4.4 **Genome**: All DNA contained in an organism or a cell, including both the DNA comprising chromosomes within the nucleus and the DNA in mitochondria.

4.5 **Database for Genotypes and Phenotypes (dbGAP)**: A central repository at the National Center for Biotechnology Information, a branch of the National Library of Medicine at the NIH.

5.0 **Responsibilities**:

5.1 **Principal Investigator** is responsible for the following:

5.1.1 Ensuring risks to study subjects are minimized by submitting data to the NIH GWAS data repository that is coded using a random, unique code, and de-identified according to the following criteria:

5.1.1.1 The identities of data subjects cannot be readily ascertained or otherwise associated with the data by the repository staff or secondary data users;

5.1.1.2 The 18 identifiers enumerated at section 45 CFR 164.514(b)(2) (the HIPAA Privacy Rule) are removed; and

5.1.1.3 The submitting institution has no actual knowledge that the remaining information could be used alone or in combination with other information to identify the subject of the data.

5.1.2 Retaining the key to the code that would link to specific individuals to the data as appropriate;

5.1.3 Ensuring The National Center for Biotechnology information (NCBI) which houses the GWAS repository never receives the code or any other information that would enable the identification of the individuals who are the source of the data;

5.1.4 Obtaining a Certificate of Confidentiality (CoC) to provide an additional safeguard with regard to compelled disclosure in any civil, criminal, administrative, legislative, or other proceeding, whether at the federal, state, or local level, of information that could be used to identify individual research subjects;

5.1.5 Submitting an application to the USF IRB when requesting data from the GWAS data repository. Research utilizing data from dbGAP is typically deemed Not Human Subjects Research (NHSR). For additional information, please see HRPP Policy 312, “Not Human Subjects Research.”

5.2 **IRB** is responsible for:

5.2.1 Reviewing the investigator’s plan for data submission, as well as the adequacy of the informed consent process and documents through which the data were obtained;
5.2.2 Ensuring the confidentiality of the data and the privacy of subjects as the genotype and phenotype information generated about individuals will be substantial and, in some instances, sensitive (such as data related to the presence of risk of developing particular diseases or conditions and information regarding family relationships or ancestry);

5.2.3 Encouraging investigators submitting data to the GWAS repository to obtain a CoC;

5.2.4 For studies submitting data and materials to the GWAS repository, the USF IRB must certify that each of the following are met:
   5.2.4.1 The data submission is consistent with all applicable laws and regulations as well as institutional policies;
   5.2.4.2 The appropriate research uses of the data and the uses that are specifically excluded by the informed consent documents are delineated;
   5.2.4.3 The identities of research subjects will not be disclosed to the NIH GWAS data repository; and
   5.2.4.4 An IRB and/or Privacy Board, as applicable, reviewed and verified that:
       5.2.4.4.1 The submission of data to the NIH GWAS data repository and subsequent sharing for research purposes are consistent with the informed consent of study subjects from whom the data were obtained;
       5.2.4.4.2 The investigator’s plan for de-identifying datasets is consistent with the standards outlined in the policy
       5.2.4.4.3 It has considered the risks to individuals, their families, and groups or populations associated with data submitted to the NIH GWAS data repository; and
       5.2.4.4.4 The genotype and phenotype data to be submitted were collected in a manner consistent with 45 CFR Part 46.

5.2.5 Documenting the appropriate certification in an approval letter for submission to the NIH GWAS data repository.

6.0 Submission of Data Collected Prospectively:

6.1 To submit data and materials to the GWAS for prospective studies (studies in which informed consent will be obtained prospectively), investigators must provide the following information to the IRB for consideration:

6.1.1 A data sharing plan which must include:
   6.1.1.1 Documentation that data submission is consistent with applicable laws and institution policy;
   6.1.1.2 The appropriate research uses of the data and any specific research exclusions as outlined in the informed consent document; and
   6.1.1.3 Confirmation that the materials and data submitted to the GWAS data repository are de-identified per the HIPAA Privacy Rule regulations and at no time will the link to the identifying information, nor the actual identifying information, be disclosed to the GWAS data repository.

6.2 The informed consent document must comply with the federal regulations contained in 45 CFR 46 and include information regarding the data sharing with the GWAS data repository.
6.3 The informed consent document must clearly state that DNA will undergo genome-wide analysis and that genotype and phenotype will be shared for research purposes with investigators who submit proposals to the GWAS data repository. (The USF Genetic Research addendum contains suggested language for use in informed consent documents).

7.0 Submission of Data Collected Previously:
7.1 To submit data and materials to the GWAS data repository for retrospective studies (studies in which informed consent was collected previously as part of a research study), investigators must provide the following information to the IRB for consideration:

7.1.1 A data sharing plan which should include:
7.1.1.1 Documentation that data submission is consistent with applicable laws and institutional policy;
7.1.1.2 The appropriate research uses of the data and specific research exclusions as outlined in the informed consent document;
7.1.1.3 Confirmation that the materials and data submitted to the GWAS data repository are de-identified per the HIPAA Privacy Rule regulations and at no time will the link to the indentifying information, nor the actual identifying information, be disclosed to the GWAS data repository.

7.2 The IRB must review the informed consent documents which were signed by subjects to confirm whether or not the initial consent under which genetic materials were obtained is consistent with the submission of data to the GWAS data repository and the sharing as outlined in the GWAS policy.

7.2.1 The IRB may determine that the original consent is not consistent with submission of data to the GWAS data repository and may request re-consent of subjects.

7.2.2 The IRB may determine that the original consent is not consistent with submission of data to the GWAS data repository and determine that it cannot verify that the criteria outlined in the GWAS policy have been met for submission of data to the GWAS data repository and therefore, such submission is not appropriate.

7.2.3 The IRB cannot waive the requirement for informed consent for the submission of data and materials to the GWAS data repository.

8.0 Withdrawal of Individual Consent:
8.1 The NIH GWAS data repository has developed policies with regard to removal of individual data records if consent is withdrawn. PIs submitting to the GWAS data repository and their institutions may request removal of data on individual subjects from the data repository in the event that a research subject withdraws consent. However, data that have already been distributed for approved research use will not be able to be retrieved.

References:
45CFR46.116
45CFR 164.514(b)(2)
NIH Points to Consider for IRBs and Institutions in their Review of Data Submission Plans for Institutional Certifications Under NIH’s Policy for Sharing of Data Obtained in NIH Supported or Conducted Genome-Wide Association Studies (GWAS) November 12, 2007.