

Privacy and Sharing of Genomic Data

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Objective: analyze human genome for diseases

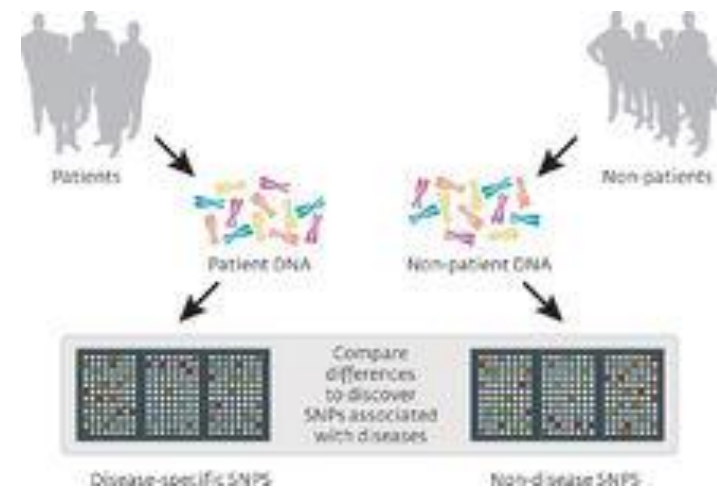
Genomic Wide-Association Studies (GWAS)

Associations between genetic variations and specific traits

- Ex.: BRCA genes and risk of breast cancer

Sharing of aggregate data

- **Simple client-server architectures**
- **Initially: no privacy problems known**



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Attacks for identification of individuals in genetic DBs with aggregate data

[Homer et al., 2008]: identification in large aggregate data sets

[Sankaraman et al., 2009]: upper bound on detection power

[Wang et al., 2009]: identification in small data sets

Result: severe restrictions to public data sharing

Ex.: **gwascentral.org**

- frequency info not available
- Large data sets only available on request
("Data Sharing Statement" of GWAS Central)

Generally: restrictions on

- **Sharing system architecture**
- **Queries on genomic DBes**



Enter a study id, dbSNP id, MeSH/HPO phenotype term, keywords, author
(e.g. HGVST307, rs2317951, Pancreatic cancer, replication st

About GWAS Central

GWAS Central provides a centralized compilation of summary level findings from genetic association studies, both large and small. We actively gather datasets from public domain projects, and encourage direct data submission from the community. [See more..](#)

Need for more advanced sharing

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Geneticists interested in more advanced sharing possibilities

Allow sharing of **larger data sets**

Enable collaborative work on **rare variants / uncommon diseases**

Advanced sharing architectures

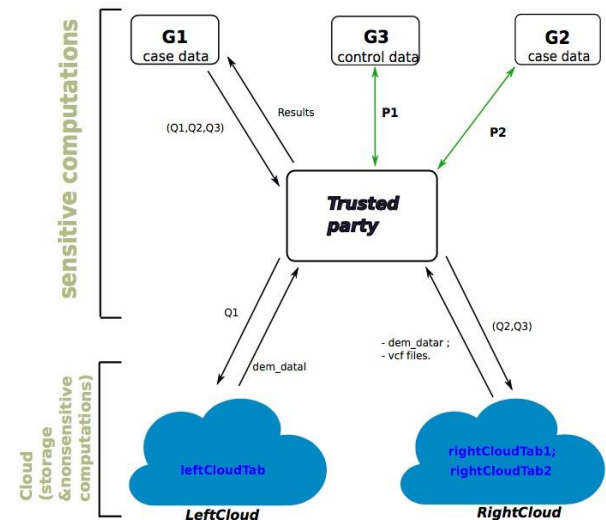
Sharing of raw data via e.g. trusted party

Quicker access to data in the cloud

Use advanced sharing techniques

Support **confidentiality** and **integrity** efficiently

Support for **ownership** and **traceability** properties



How to support such sharing scenarios?

Methods for the construction of architectures/processes/queries

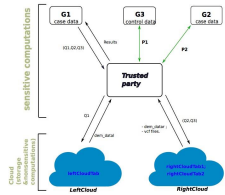
- Means for **design** and **programming**
- Ensure basic **privacy guarantees**
- **Optimize** architectures/applications
- **Formal verification** of advanced privacy properties

How to ensure privacy properties?

Multiple privacy enhancing techniques:

- **Encryption**: a/symmetric, homomorphic, attribute-based, ...
- **Client-side computing**: compute associations within local perimeter
- **Data fragmentation**

- **Watermarking** for ownership and traceability properties



Declarative scenario definitions with privacy types

```

scenario : GeneticQuery [SubjectId, ZIP, Gender, DoB, Variant, TypeVar, MyTattoo]
scenario = do
  G1 'SendRequest' (TTP, [Q1])
  G1 'SendRequest' (TTP, [Q2, Q2'])
  G1 'SendRequest' (TTP, [Q3, Q3'])

  TTP 'SendRequest' (LeftCloud, [Q1])
  TTP 'SendRequest' (RightCloud, [Q2, Q2'])
  TTP 'SendRequest' (RightCloud, [Q3, Q3'])

  let q1 = LeftCloud 'executeRequest' [Q1];
  let q2 = RightCloud 'executeRequest' [Q2, Q2'];
  let q3 = RightCloud 'executeRequest' [Q3, Q3'];

  demData ← LeftCloud 'SendData' (TTP, q1)
  demDatar ← RightCloud 'SendData' (TTP, q2)
  vcfFiles ← RightCloud 'SendData' (TTP, q3)

  let r1 = decrypt VariantWE (AESD "key2") vcfFiles;
  let r2 = decrypt TypeVarWE (AESD "key1") r1;
  let r3 =
    detectw VariantW (RGIG "wkey1" 1 ["seed1"] 1) r2;
  let vcfFiles =
    detectw TypeVarW (RGIG "wkey2" 2 ["seed2"] 2) r3;
  let plainData =
    defrag (defrag demData demDatar) vcfFiles

  TTP 'ReturnResults' (G1, TTP 'Compute' plainData)
  
```

```

Wat : (a : Attribute) →
  {auto p1 : So (isRawType (snd a))} →
  {auto p2 : So (isInEnv a env)} →
  Privy env (watEnv a GIG env) []

— watermark detection operator

data Query :
  ...

detectw :
  (a : Attribute) → (info : ReadM GIG) →
  {default Refl p1 : (snd a) = (WATERMARK GIG t)}
  → Query Δ →
  {auto p2 : Elem a Δ} →
  Query ((replaceOn a (fst a, t) Δ)++[MyTattoo])
  
```

Definition of watermarking operator

Algebraic theory: watermarking laws

$$\begin{aligned}
 & \text{decrypt}_{(s,a)} \circ \text{crypt}_{(s,a)} \circ \text{detectw}_a \circ \text{wat}_a \equiv \\
 & \text{detectw}_a \circ \text{decrypt}_{(s,a)} \circ \text{crypt}_{(s,a)} \circ \text{wat}_a \\
 & \text{if } \text{dom}(p) \cap a = \emptyset \\
 & \text{detectw}_a \circ \sigma_p = \sigma_p \circ \text{detectw}_a
 \end{aligned}$$

Verific./Optim. of genetic applications for privacy/efficiency

$\pi_{(variant, typeVar)} \circ$

$\sigma_{((subjectId \in mdd) \wedge (position=i, position=j, \dots))}$

(a) local query

$\text{detectw}_{variant, typeVar} \circ \text{decrypt}_{variant, typeVar} \circ$

$\pi_{(variant, typeVar)} \circ$

$\sigma_{((subjectId \in mdd) \wedge (position=i, position=j, \dots))} \circ$

$\text{crypt}_{variant, typeVar} \circ \text{wat}_{variant, typeVar}$

(b) distributed query

- **How to harness/integrate other PETs (e.g., differential privacy)**
- **Which kind of genetic data and analyses can be safely outsourced?**
- **What about new analyses?**