Crowdsourcing Genome Wide Association Studies

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Overview

What are GWAS?

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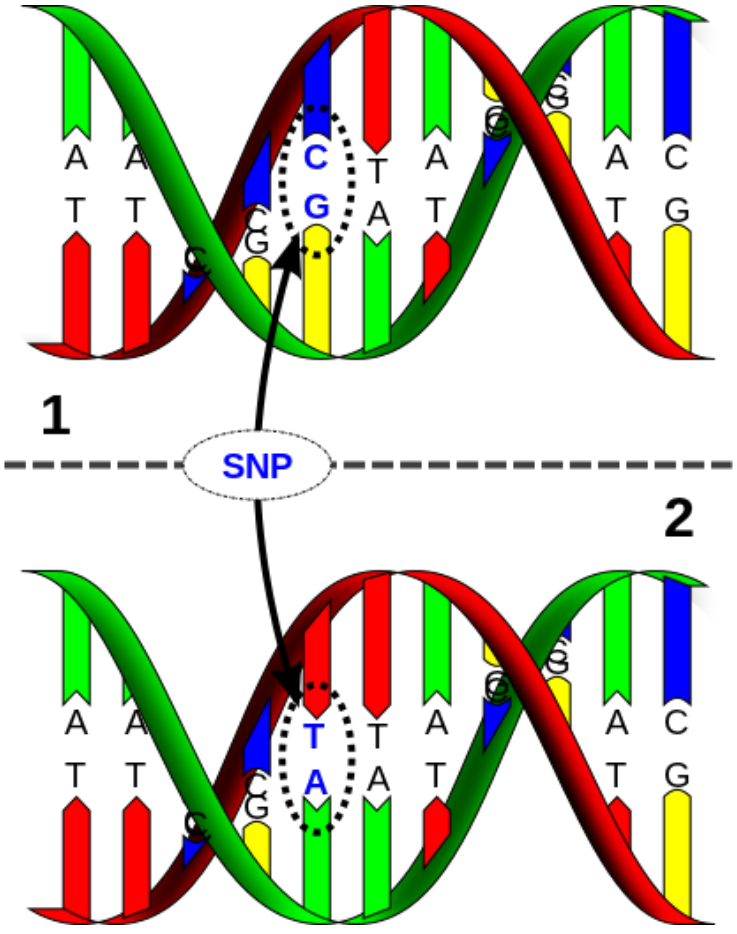
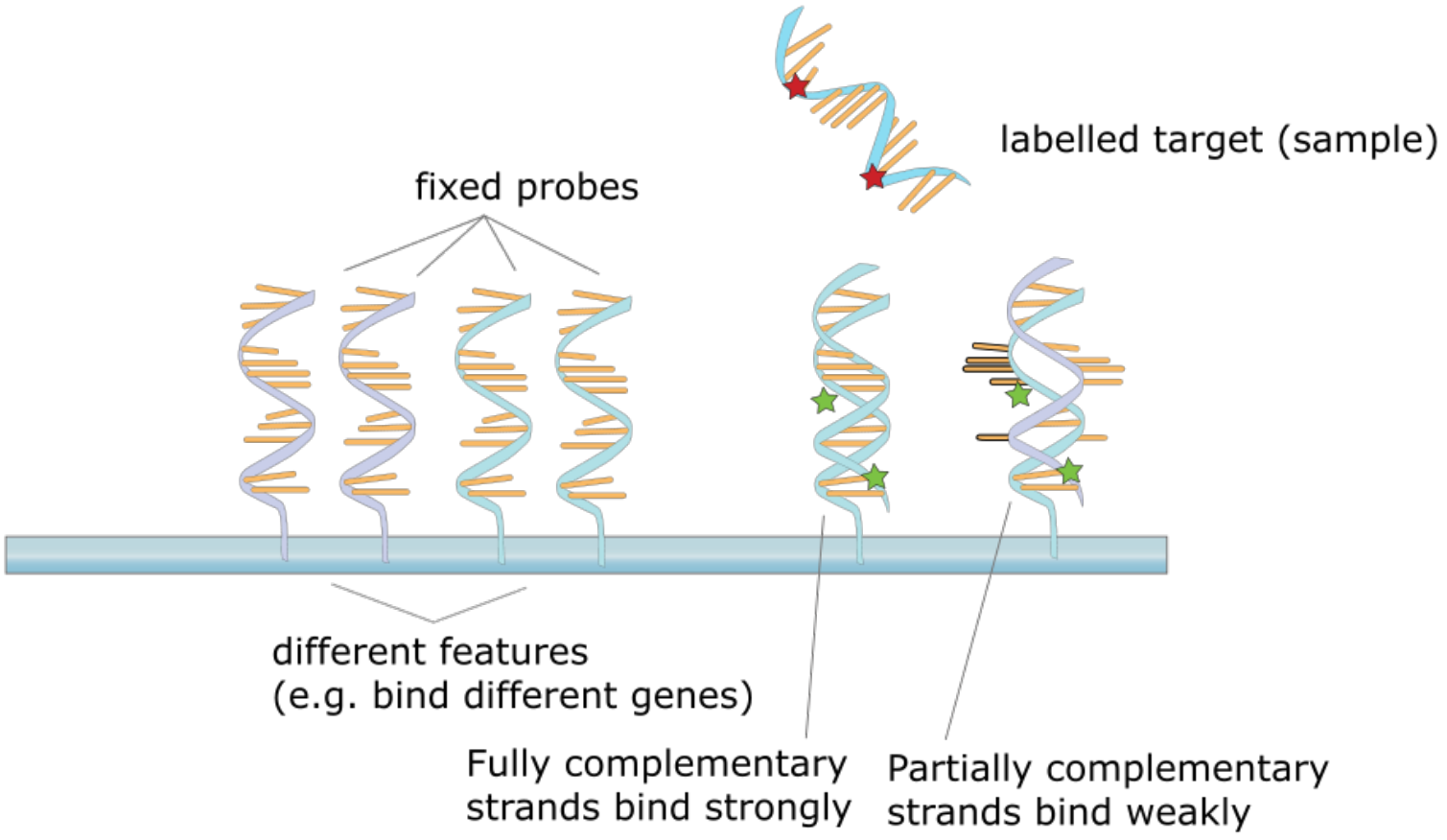
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like Diabetes
- Compares groups carrying a certain SNP with groups without that SNP

Single Nucleotide Polymorphism



Source: http://en.wikipedia.org/wiki/File:Dna-SNP.svg

How do GWAS work?



Source: http://en.wikipedia.org/wiki/File:NA_hybrid.svg

Some GWAS-examples

- Sladek *et al.* (2007) identified four gene locations linked to heightened type 2 diabetes risk

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- Kogan *et al.* (2011) linked rs53576 (G:G) to pro-social behaviour
- The Wellcome Trust Case Control Consortium (2007) linked 24 locations to 7 major diseases

Personalised GWAS

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- You get access to the raw data!

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- No way for scientists to access the data
- Some customers uploaded their data to the net

What can happen with open data?

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- Positive and negative consequences
- Possibly extremely bad consequences
- Up to you to decide whether you want to open your data

Positive consequences

- More knowledge about yourself

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- Cheap, open science

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- More knowledge about yourself
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- Great data-source for citizen scientists

Negative consequences

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- People know more about you than you'd like
- Your boss, your insurance company...
- Personal SNPs very similar to parents and relatives
- You could be carrying a deadly disease

Why you should care

- It's time to take science into our own hands

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- It's time to take science into our own hands
- You can make this happen!

Open GWAS

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- So far: 78 genotypings and 188 users

Conclusions

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- Open GWAS are the future of personalised medicine
- It's in the hands of users to make or break the situation

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- Constantly improving the project
- Trying to get funds for free genotypings

The end

Thanks for listening. Any questions?

References

Kogan, *et al.* (2011): Thin-slicing study of the oxytocin receptor (OXTR) gene and the evaluation and expression of the prosocial disposition. Proceedings of the National Academy of Sciences
Sladek *et al.* (2007): "A genome-wide association study identifies novel risk loci for type 2 diabetes". Nature 445 (7130): 881-5.
The Wellcome Trust Case Control Consortium (2007): Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature 447: 661-678.