SDgeneMatch, a new tool to aid the identification of the genetic causes of DSD

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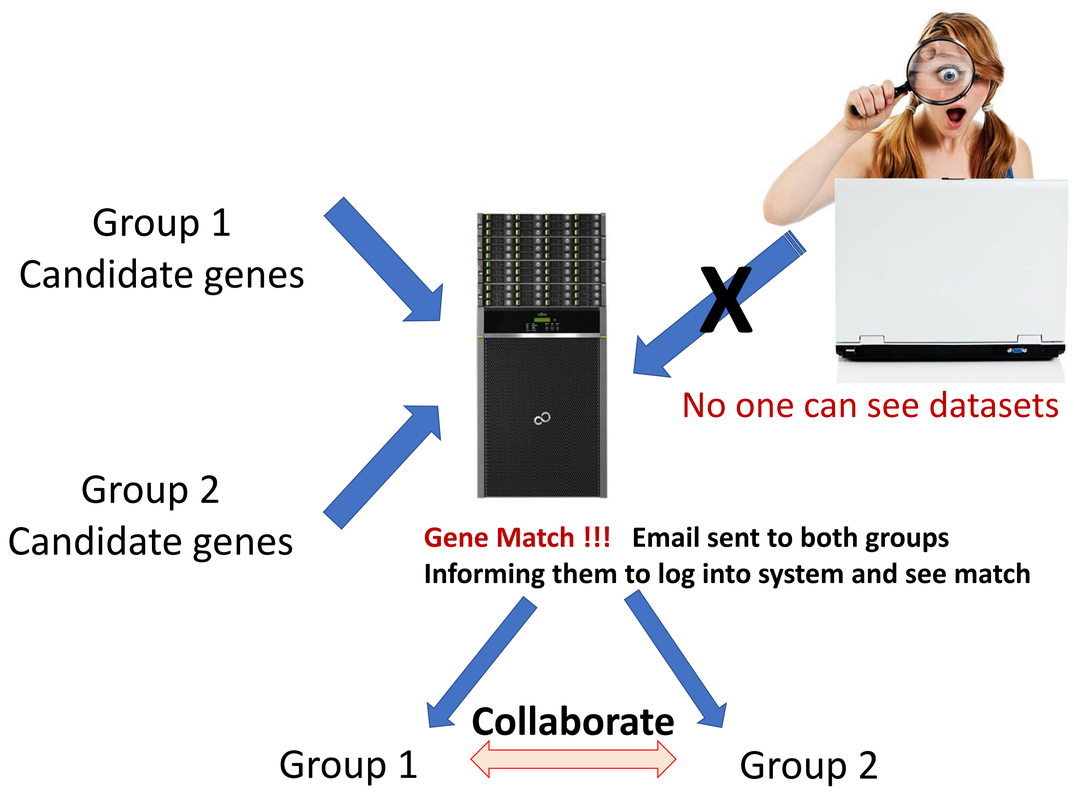
Background: Genomic analysis is slowly revealing new genetic causes of DSD including *SOX8*, *ZNRF3* and *NR2F2* (1-3), atypical DSD presentations associated with mutations in known DSD genes (4) or evidence for digenic causes of DSD (5). As high-throughput sequencing of DNA from patients becomes more widespread, there is an increasing number of candidate new DSD genes. The interpretation of datasets from rare diseases, such as many forms of DSD, is complex since all human genomes carry many deleterious mutations. Analysis of datasets can lead to false associations between mutations and phenotypes unless it is possible to provide statistically compelling evidence in favour of a gene mutation being causal.

As part of the COST Action BM1303 A Systematic Elucidation on Differences of Sex Development (www.dsdnet.eu). We decided to develop a genomic datasharing platform termed SDgeneMatch. The aim of this is to share between interested clinical and research groups rare and novel genetic variants that potentially could contribute to DSD, whilst maintaining complete confidentiality of the genomic dataset.



Sharing DSD NGS Data - SDgeneMatch

NOT – Data Analysis
NOT – Data storage

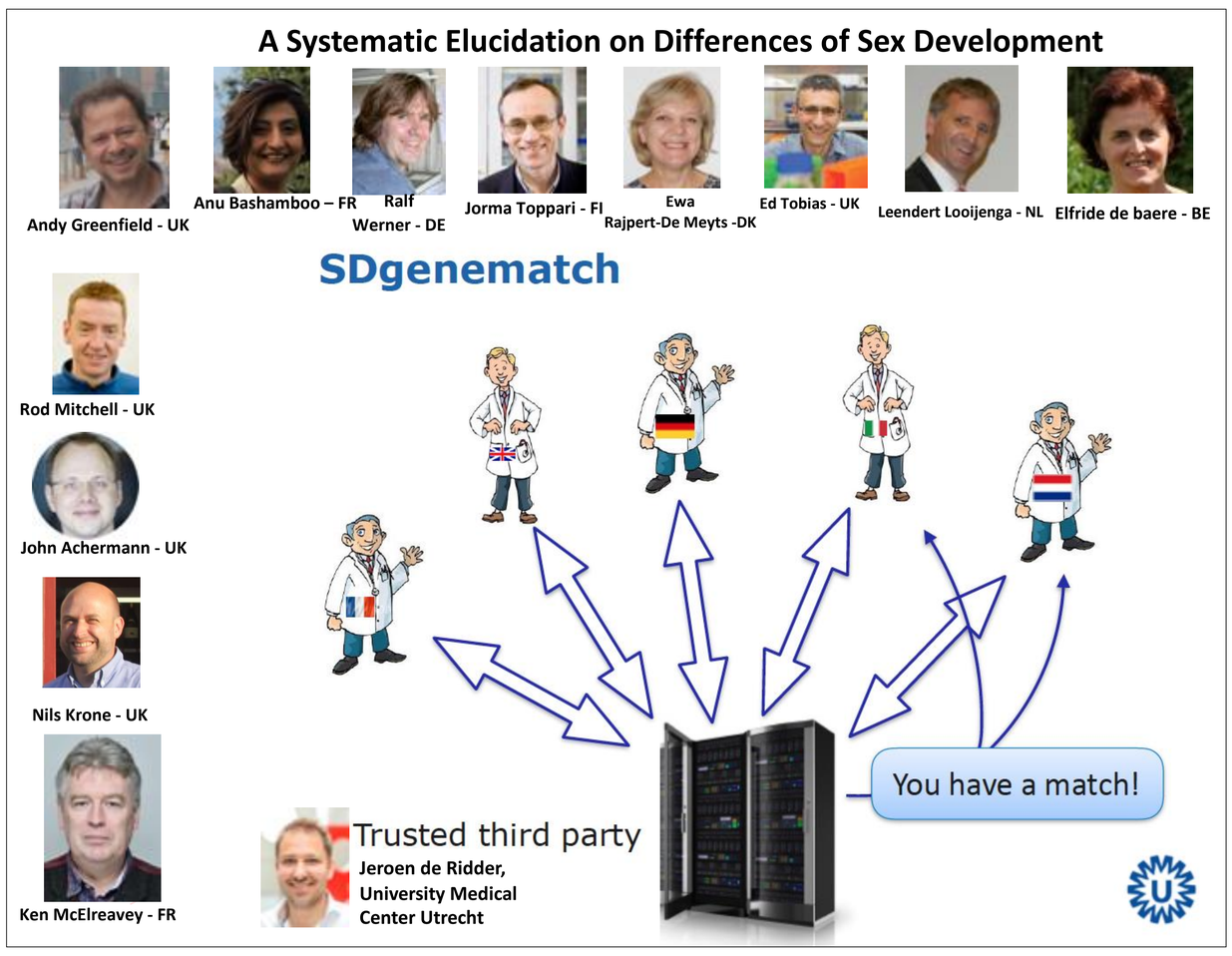
Simply matching genes between groups

Very simple procedure –

Add gene name(s) only into the system

-- No details of the phenotype
-- No details of the mutation(s) themselves
-- No other information introduced into the system

No one can see your information introduced into the system



Members of the COST Action WG2 and the development of the SDgeneMatch system

The database now contains information on more than 500 exomes. Data has been submitted from various European and Australian groups and it is now open to anyone generating datasets from DSD patients. This should provide a valuable tool in the identification of new genetic causes of DSD.

References:

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