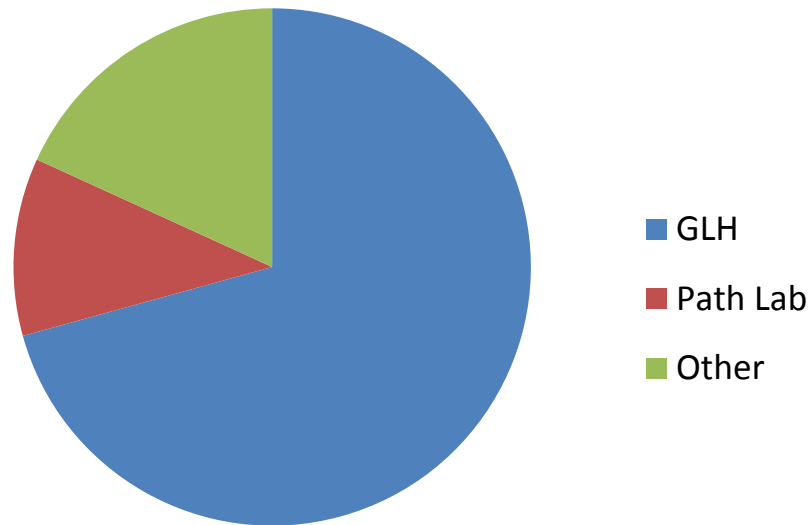


Transition to build 38 survey

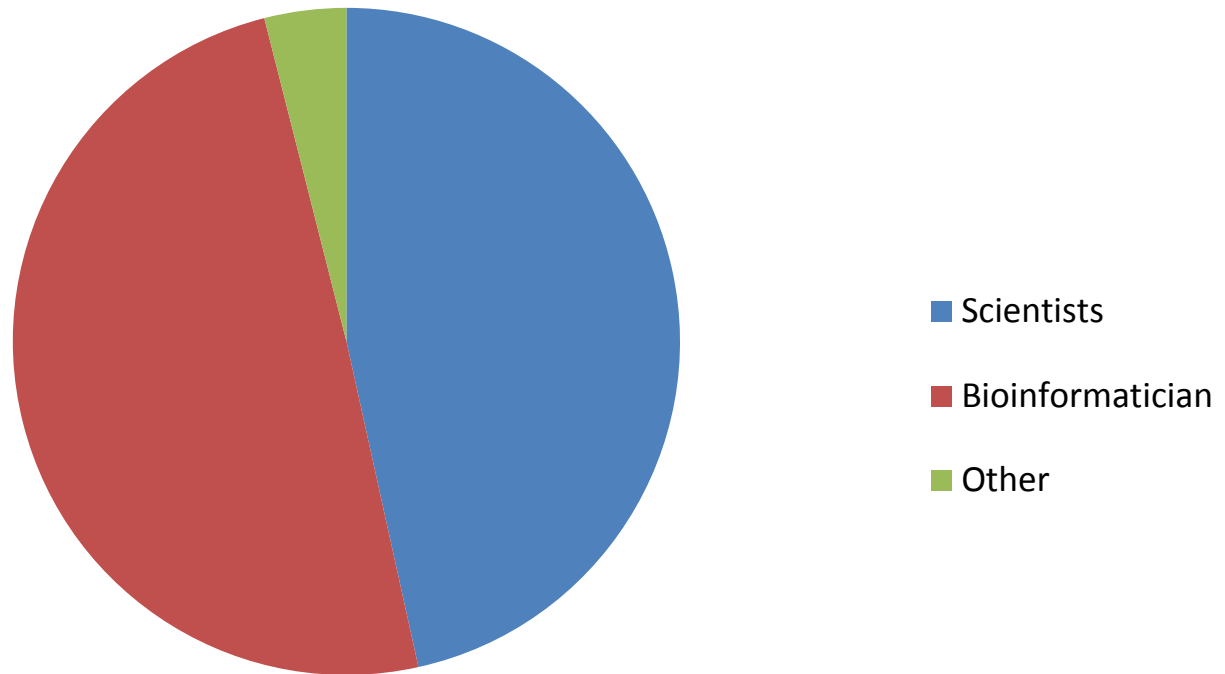
# Where?

- 44 replies – only 28 actual responses



- Other - Regional Genetics Scotland; Clinical Genetics; siHMDS; Academic lab; Path lab ex UK; Gov funded lab; Charity

# Who?



# Who is actively planning to move to build 38 in 2019?

- No 50%
- Yes 50%

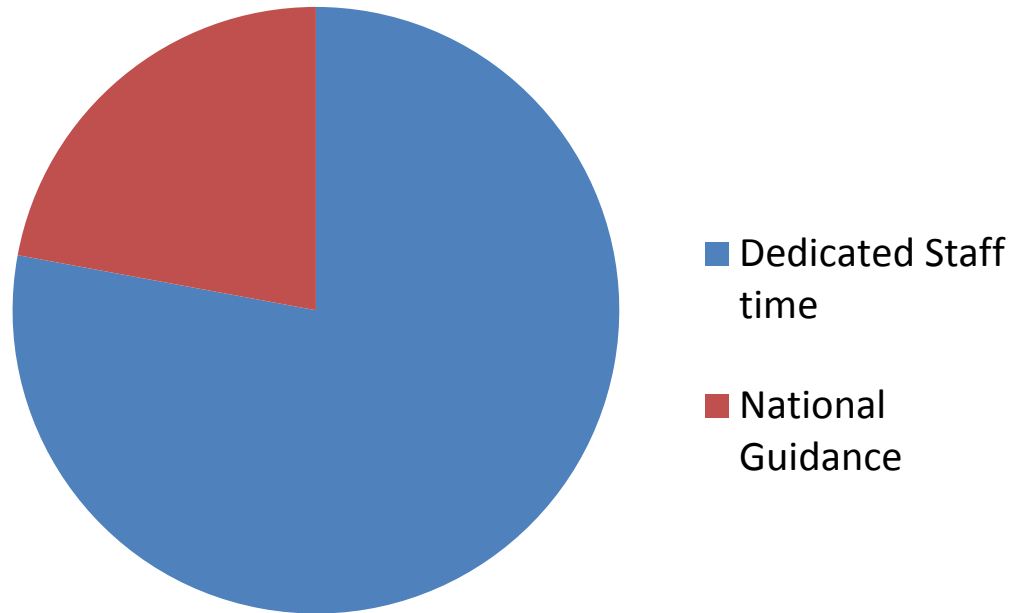
# Major Obstacles 1

- Reference databases eg CNV interpretation databases (e.g. DECIPHER); **Gnomad**; Cancer Genome Interpreter
- Publications
- In-house legacy data is in build 37
- UK network of labs need to all be using same numbering to avoid confusion when sharing data.
- Time constraints, lack of staff time to validate
- IT

# Major Obstacles 2

- External providers of NGS analysis / variant calling still using build 37
- Compute resource to run validations of all the NGS panels
- HGVS nomenclature
- Pipeline development, annotation database updating and data consistency of trials
- Uncertainty about the need to make the move- i.e. why should we bother? Fear of the unknown
- Easy re-numbering of variants detected in hg37 to hg38

# What resources would be required?



## Comments

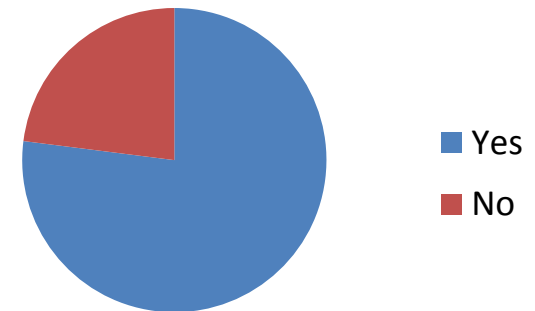
- Commercial software needs to move
- Bioinformatics taking the lead

# Are Existing Resources Compatible?

Compatible	Not Compatible
Alamut	CNV interpretation database
Array analysis software	Sophia genetics
ANNOVAR/InterVar	Thermofisher Ion reporter
VEP	NGS analysis software from commercial provider
Mutalyzer	In house pipelines
	GnoMad?
	Cancer genome interpreter



# Do you see any issues around local vs national data?



- Comments

- Need conversion step for build 37 – could be confusion which data needs conversion
- Could be problems storing build 38 100KGP data in local build 37 database
- ? submitting build 37 to a national build 38 database
- IT infrastructure
- Backwards compatibility- eg requests to reanalyse NGS data from last year. Would need to maintain some level of support for GRCh37 to be ongoing.
- confusion if some have moved others have not

# Discussion

- External databases/software
- Legacy data mapping
- Working with both builds
- Cancer vs Rare disease
- Why, How, When?

# ACGS Build 38 Meeting

Room 1

Friday 17<sup>th</sup> Jan