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Subject: Notes from the runs discussion
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Notes on experimental plans for sequencing projects:

Where are we on various projects:

Ops:

- a. Get technical baseline and get data with modern seq biochem
 - i. Standard WGS trio
 - 1. Sequencing done – amy good, father and son 2nd read ”passed” but not great.
 - 2. Working through issues in analysis.
 - 3. Can look at things like coverage, GC bias, other non-familial parameters.
 - 4. Shown we can get good data, but may not every time.
 - 5. Suspecting reagent issues.
 - 6. Do we need additional baseline work? Yes. We’re still working out our processes so we’re getting the same thing consistently. Amy runs more useful than phiX runs.
 - 7. Can continue with project data and still look at trends in the data.
 - 8. Need run tracking metrics consolidated for the runs that have been completed. (Rose)
 - ii. Standard Exome:
 - 1. Running 24 (CEPH family + some others)
 - 2. We have half a run – R2 issues, but R1 looked good.
 - 3. Rerun starts 10/4 - 2x100 run (12 days)
 - iii. 2nd HiSEQ

1. finishing qualifying run.
- iv. Storage size – 30TB
 1. Can park data on HDs rather than deleting if need be.
- v. R&D objective for accuracy –
 1. get data with modern sequencing biochem (aka V3)
 2. profiling of one sample (AMY). Value is beginning to drop off.
 3. additional goal is a cross samples for concordance (across diversity panel?)
 4. duplicate library preps and pullouts
 5. could run with Moyamoya samples? Should have enough for 2 or 3 attempts on each sample.
 6. Mark make a plan for replicates on this.
- vi. OPS objective for standardizing processes and getting lab up and running.
- b. CLIA validation -
 - i. Need CLS person on-board before we can do validation
 - ii. 24 CFTR samples – DNA is in house.
 - iii. Only 2-3 libraries so far though.
 - iv. Need clarification on whether we need to be CLIA for VA project. If nothing, we plan to pursue CFTR approach. First item to be approved on would be standard exome seq for the purpose of diagnosing CFTR.
 - v. WGS of samples useful later for WG addition to our menu, but not on the to do now.
 - vi. WGS of NIST aliquot – NA12878 for proficiency testing – unclear about consents.
 - vii. CLIA validation in Nov.

Customer projects:

- a. Moyamoya 132 exomes project
 - i. Can be used for Mark's coverage statistics.
 - ii. MiSEQ run finished and looks good with test samples (one pool of six). On/off target ratios look good.

- iii. 1st Moyamoya run starts today (10/4) – 24 samples.
 - iv. 2nd library prep under way – start run
 - v. Thursday next week (10/11), 2nd Moyamoya run can start. -second batch of 48.
 - vi. Would have 72 done in 3 weeks.(10/22)
 - vii. On 10/22 start 48 more. – need to see if we can pull in library prep.
 - viii. 10/25 could start final group plus any reruns and some replicates (for understanding variability).
- b. Moyamoya familial collection – additional samples from Stanford.
 - i. Not until 132 exome project completed.
 - c. VA 1M veterans project

R&D projects:

- a. Testing pullouts for EXOME+, WGS+, Panels
 - i. Need to figure out when to slot in experimental plan.
- b. Determining accuracy dependence on seq ops parameters
- c. Samples with known pathogenic STR variants