ABSTRACT
Motivation: Our goal was to combine the capabilities of Spark and GOR into a single computing framework for use in analysis of large scale genome data.

Results: We have created a relational query engine that unites SparkSQL and GORpipe into a single declarative query framework. This has been achieved by allowing embedding of SQL expressions into the high-level relational statement syntax in GOR and by supporting virtual relations and nested GORpipe expressions within SQL. Furthermore, we have built drivers to enable Spark and GOR to use and leverage their preferred file formats, Parquet and GORZ respectively, and introduced APIs to allow the use of GOR with Spark dataframes.

Availability: The SparkGOR version of the GORpipe software is open-source and freely available at https://gorpipe-website.now.sh and https://github.com/gorpipe.

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1 INTRODUCTION
Like many other domains, genetics has for a while been a field of big data analysis, because of the general availability of high-throughput sequencing technology for data generation and because of continuous improvements in data storage and computing capabilities for data analysis. As data generation keeps growing at an increasing rate and there is a desire to analyze ever larger cohorts of samples, it continues to be important to improve and optimize the software analysis systems.

We released the GORpipe system few years back with them aim to provide an efficient general purpose analysis tool based on a genome ordered relational data architecture and declarative pipe-syntex query language [7]. The GORpipe system does provide a rich set of commands to work with sequence read variants and segment based data for use in rare-disease analysis as well as large scale cohort analysis. For instance, it is being used as the database for the genomics learning system in the Children’s Rare Disease Cohorts initiative (CRDC) at Boston Children’s Hospital[13]. Our aim here is to open-source and integrate the GOR system into Apache Spark, a popular unified general purpose computing platform for big data analysis [19].

The initial Spark architecture is based on resilient distributed data sets (RDDs) that have APIs that lend themselves elegantly to functional style programming. Because of its memory based caching architecture, it has been shown to outperform Hadoop by orders of magnitude, especially in iterative machine learning algorithms and interactive analysis. Although the general RDDs can be used to process semi-structured data, as in the MapReduce paradigm [3], much of the focus in past years has been around structured data, loosely typed relational DataFrames and DataSets with strictly typed object bindings, and SparkSQL which is arguably one of the most important and powerful feature in Spark. While the fundamental architecture of Spark has changed little over the years, the implementation has undergone significant changes between major releases. As an example are feature improvements introduced with project Tungsten, which was driven by focus on structured data and the fact that CPU had become more of a bottleneck than IO and network bandwidth. These features include off-heap memory management for reducing JVM garbage collection overhead, improvements in object serialization, cache aware data structures, code generation, and vectorized computation, leveraging columnar data formats and low level parallel instruction sets.

There are many similarities between the two platforms, GOR and Spark. Both use distributed JVM execution engine for running parallel declarative queries, both have the capabilities to access read only tabular data from various file formats, and both have a joint focus on interactive and batch processing analysis. There are also significant differences; unlike Spark that implements off-heap memory based serialization for caching of data, GOR relies on the file system client caching mechanism (e.g. NFS client caching) but caches only meta-data explicitly, such as file partition and genome positional information. The GORpipe query language supports views and syntax to refer to federated virtual relations, that may represent federated or client based data. The GOR system automatically re-evaluates such virtual relations and materializes them to a file cache using logic based on data time-stamps and expression signatures. Spark however requires explicit registration of temp view (e.g. df.createOrReplaceTempView) but uses lazy evaluation and optimizes transparently task scheduling, taking into consideration optimal memory data caching. Spark supports both SQL query style usage as well as SDK-API for functional and imperative style programming, in an interactive shell or in batch mode. Sofar, the GORpipe system has only support for multi-step pipe-query analysis and we have not yet provided APIs aimed for SDK bindings, something we change with the Spark integration presented in this work.

The primary difference between these two systems is however their standard execution approach for joins and parallel processing. GOR relies on ordered data structures and tries to optimize the use of random access vs full-scan access in merge-hash joins. For controlling parallelism it uses genome coordinates and seeks to split tasks along the genome axis, while it can also parallelize tasks along the data partition axis1. Results from different query steps and tasks are communicated through a shared file system. Currently, for both

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E.g. samples for genotypes and phenotypes for GWAS results.
types of parallelism, it must be instructed explicitly through the syntax, e.g. via PGOR or PARALLEL commands. Spark however relies primarily on full-scan of data and uses broadcast or shuffle based hash-joins or sort-merge-joins (which typically also uses shuffle operations), analogous to how many SQL engines have been implemented in the past, such as those who are Hadoop based. SparkSQL also has Adaptive Query Execution (AQE), an optimization technique that uses runtime statistics to choose the most efficient query execution plan. Users can also control the level of parallelism in a more fine grained manner by using hints in SQL or through the SDK-API.

Several projects have used or extended Spark to work with genome data, such as ADAM [10], GATK4 [12], GenAp [9], Hail [17] and Glow [16]. The early versions of ADAM focus on secondary analysis and the use of well formed AVRO schema and Parquet columnar storage to represent sequence read and variation data. Later, they have also extended the Parquet format with metadata to better support efficient range lookups and implemented broadcast and shuffle range join logic. In GenAp, a RANGEJOIN extension to SparkSQL is introduced, similar to the high level syntax in GQL [8] and joins of pipe expressions in GORpipe. They use interval tree approach to enhance a broadcast join, to speed up the interval joins of genome features, whereas GORpipe relies on pre-ordered data and seek-scan merge join logic. In Hail the focus is on cohort analysis, kind of a Spark driven Flink system [14]. They recognize that sample based row-level representation of genotype data can create large overhead in statistical cohort analysis. They introduce Matrix tables that are similar in nature to annotated BGEN and pVCF files, implementing methods for lazy distributive analysis in Spark SDK. Glow focuses also on distributed statistical cohort analysis, however, they use standard Spark DataFrames to represent cohort genotypes, storing them in regular array-based value column that Parquet supports. This is analogous to the approach used in GOR for representing horizontal layouts, although the commands in GOR also allow data such as genotypes to be split into multiple buckets, each bucket using a separate value row for each variant. This is similar to the approach used in GenomeDB [2][13], allowing for easier updates and partitioned read access. GORpipe does also have commands that efficiently use such bucketized horizontal value format representation, for Fisher-exact test, logistic regression, LD calculation, relationship analysis, and subsetting or transposing the data back to row-level representation.

Here we present SparkGOR, our work on integrating GOR with the Spark architecture, including the following items:

• We present a relational query engine (RQE) that integrates SparkSQL and the GORpipe syntax into a generic relational query language.

• Enable parallel execution of GOR queries within the distributed Spark execution environment by building Spark-RDD query handlers for GOR.

• Extend the DataFrame API with methods for GOR, allowing the GOR expressions to be used with Spark SDK.

• Develop drivers and adapters allowing Spark and GOR to leverage each others preferred data formats and partition strategies, Parquet and GORD/NOR respectively.

In the following sections, we describe the RQE, the unified relational query language, by showing selected examples that explain its nature and the motivation for our integration. Then we present the SDK integration with few simple Scala code snippets that show how GOR can be used with Spark DataFrames as well as an extract from a more extensive PySpark script based on GloWGR [11], that is an excellent candidate for an analysis workflow deployed with Spark Kubernetes Operator.

2 RELATIONAL QUERY ENGINE

To understand the extension of GORpipe to RQE requires a basic knowledge of the GORpipe syntax described in [7] and [4]. The formal structure is zero or more CREATE statements followed by a single GOR or NOR expression that returns the final relation. This relation can be based on one or more relations defined with the previous create steps. The reference to the create statements forms a directed acyclic graph which determines the execution order and re-evaluation dependencies. In standard GORpipe syntax the expressions start with GOR or NOR to determine ordered or non-ordered execution context. Here, in SparGOR, we add expressions that start with a SELECT command, indicating SparkSQL context and the Spark execution engine2. Unlike in regular SparkSQL expressions, that query relational data sources and files that are registered into the sqlContext, we additionally allow these data sources to be materialized virtual relations (...), nested GOR/NOR expressions <(...), and a tail-expression separated with pipes, e.g. | pipe-step(s).

As in previous work on GORpipe, rather than providing a formal syntax specification, we believe it is more informative to explain this with selected examples. This is especially so, because RQE does not rely on a formal syntax specification for each sub-query engine. Rather, it carves out the virtual relations and nested expressions and injects the necessary data sources appropriately. The benefits of this injection approach is that it can for instance leverage all the context specific user-defined SQL functions and is also insensitive to changes in the SQL syntax specification.

To illustrate the above, first consider the following simple case:

Example 1: Create statement based on SparkSQL

create #r1# = select * from genes.gorz
where chrom = 'chr1';
nor [#r1#]

There are few things to notice here. First, that in the SQL expression the filename genes.gorz can be cited directly, like in GOR and NOR expressions. Second, we have built drivers to allow the SparkSQL engine to processes GORD and the block compressed GORZ file format, with seek option based on predicate push-down from genome range filter, as shown above. Thus, the read execution is quite similar as in gor -p chr1 genes.gorz, however, unlike the materialized outputs of GOR and NOR, that are stored in GORZ and TSV respectively, the output of a SparkSQL query, such as [#$r1#$], is in the Parquet format which is now seamlessly read by the NOR command.

Now consider a case where a user wants to identify variants based on some rsIDs of choice, e.g. to find annotation information on allele

2 We have also designed a more explicit and generic syntax that supports loosely coupled federated execution engines, as compared to the implicit and tight integration between Spark and GOR as described here. Description of it is however outside the scope of this work.
The SparkSQL engine will then execute the query in parallel using its preferred strategy. Sorting 700 million rows may take a while, however, in the context of our example this is a one-time execution, since it is automatically cached to files by GORpipe and is independent from the rsIDs that are of interest to the end-user (e.g. rsIDs starting with 'rs222'). Only if the file dbsnp.gorz changes will it be re-evaluated.

Importantly, the query behind #myrssnps# runs in sub-second time because the predicate push-down logic in SparkSQL can take advantage of columnar range metadata associated with row groups in the Parquet file format. Another important thing worth noting in Ex. (3) is the #myordrssnps# statement and the need for ordering the data. The PGOR and the VARJOIN commands in #myphewas# require their data sources to be ordered. Rather than performing the ordering in GOR with SORT genome, we could have added an ORDER BY command into the SQL statement that does the rsID filtering, in order to ensure that the Parquet file is ordered. Similarly, we could have pre-generated an ordered Parquet file and written Ex. 3 in the following manner:

Example 4: Alternative version of Example 3

create #myordrssnps#
= select * from dbsnp.rsOrd.parquet
where rsID like 'rs222%' order by chrom, pos;
create #myphewas#
= pgor [myordrssnps] | varjoin -l -r PheWas.gord;
nor [myphewas] | sort -c pval:n,rsID

In the above formulation, the PGOR command will seamlessly add chromosome range filters on the Parquet partition files stored in #myordrssnps# and generate an ordered merge to preserve the genome order of the input to the VARJOIN command. The number of partitions will be determined by the SparkSQL logic. It is not trivial to compare the performance gains from the parallel sort in Ex. 4 because range based access of #myordrssnps# is slightly faster for GORZ files, which is the output in Ex. 3. For most practical cases, the speed difference will be minor. Finally, it is worth noting that it is important to sort the dbsnp Parquet file based on rsIDs in order to achieve a lookup time in a second or less. As an example, if the 700 million row file is ordered by genome location, the rsID filtering typically takes between 15 and 90 seconds, depending on cluster size and the caching state of the table.

Before we end this section, we want to give a short example using a tail-expression that can be used to bundle functionality only available in GOR into SparkSQL:

Example 5: DAG based filter of phenotypes

create #mySubjects#
= select PID, hpo_code from phenotypes.parquet
where year(date) > 2000
order by PID
| where hpo_code indag(hpo_parent_child.tsv,'HP:0001507')
| group -go PID -lis -sc hpo_code
| rename lis_hpo_code HPO_codes;
gor exons.gorz | where gene_symbol = 'BRCA2'
| join -sagsnps -f 20 -ir
< gor variants.gorz -ff #mySubjects#}
In the above example, we filter HPO based phenotypes using the HPO ontology DAG, stored in `hpo_parent_child.tsv`. This requires a SQL flavor with `connect-by` or recursive SQL, neither of which is available in SparkSQL today. A special `INDAG` operator is however available in GOR, allowing us to filter on the HPO term `HP:0001507` or any narrower definition. The `tail` operator is however available in GOR, allowing us to filter on `PID` thus we must be careful and order the results by `GROUP` command, to list the HPO codes in one row per subject, in parallel by the SparkSQL engine. Here we are also applying a `GROUP` command, to list the HPO codes in one row per subject, thus we must be careful and order the results by `PID`. This is to ensure that the GOR grouping command "sees" all the phenotypes for each subject when the SparkSQL engine distributes the query in parallel. Currently, this is not automatically handled by our injection logic.

Alternatively, we can also use a special `DAGMAP` command and expand our high-level HPO term of interest into all the leave nodes, the `dag_node` column, and generate a small relation `#myHPOs#` with all the HPO codes, that can then be joined with the phenotype table using SQL as shown below:

**Example 6:** Alternate version of DAG based filter

```scala
create #myHPOs#
  = nor hpo_parent_child.tsv | where child in ('HP:0001507')
  | dagmap -c child hpo_parent_child.tsv
  | select dag_node | distinct
  | rename dag_node code;

create #mySubject# = select pheno.PID as PID,
  concat_ws(',',collect_list(hpo.code)) as hpo_codes
  from phenotypes.parquet pheno
  join #myHPOs# hpo
  on pheno.hpo_code = hpo.code
  where year(date) > 2000;
gor exons.gorz |... as before
```

The above formulation of the query is fully equivalent to Ex. 5, however, pushes more work into the SparkSQL engine, taking greater advantage of parallelism and predicate push-down logic.

The above shows the flexibility of being able to mix seamlessly together GORpipe queries with SparkSQL, taking advantage of the strengths of each language, execution model and data formats.

### 3 SPARKGOR SDK

Relational query languages, like SQL and the GORpipe syntax, provide an elegant declarative approach to analyse data in an efficient way. However, there are limits to that approach and it may for instance be impossible to express some iterative conditional analysis, as is possible with imperative programming languages. Interactive shells for languages like Scala and Python, integrated into notebook environments like Jupiter or Zeppelin, provide a very elegant framework for distributed data analysts with Spark through the use of RDDs and DataFrames. This allows users to mix together SQL expressions with functional and imperative programming style, enabling them to extend the standard capabilities, taking advantage of lambda functions, closures and loops. This also enables the definition of complex multi-stage workflows that return multiple outputs, something which is often difficult or impossible to express in a single GORpipe query script, thus requiring embedding GORpipe into something like bash-shell or Nextflow scripts.

In SparkGOR, we provide a library that primarily does two things: provide a constructor for SparkGOR session that can generate Spark DataFrames from GORpipe expressions, and implicit Scala methods to apply tail-expressions on generic DataFrames. Again, we think this is best explained with few simple examples:

**Example 7:** Basic SDK usage in Scala

```scala
import org.gorpipe.spark.SparkGOR
import spark.implicits._
val sgs = SparkGOR.createSession(spark,"gorconfig.txt")
val myGenes = List(‘BRCA1’,’BRCA2’).toDF("gene")
myGenes.createOrReplaceTempView("myGenes")
sgs.setCreateAndDefs("create #mygenes# = select gene
  from myGenes;
def #exons# = exons.gorz;
def #dbsnp# = dbsnp.gorz")
sgs.setCreate("#myexons#",
  "gor #exons#
  | inset -c gene_symbol [#mygenes#]"
  val exonSnps : DataFrame = sgs.dataFrame("pgor [#myexons#]
  | join -segsnp -ir #dbsnp#
  | join -snpsnp -r #genes#")
val snpCount = exonSnps.groupby("gene_symbol").count()
```

The above example demonstrates how SparkGOR is added into a Scala/Spark shell with a simple import of our gorpipe JAR. A SparkGOR factory object, that is provided a reference to the spark session and GOR config file, is then used to create one or more session objects, e.g. `sgs`. In Scala, through the use of implicits, we can easily convert a list of genes to a dataframe and associate it to the SQL session as a temporary view, e.g. `myGenes`. Through the SparkGOR session object, we can then specify `CREATE` and `DEF` statements that become "visible" in our dataframe definition, as in `exonSnps`. Here we show two methods to do this; one that initializes multiple creates and definitions through one invocation and one that incrementally adds one create to the session, `sgs`.

Note that the dataframe definition is lazy and the PGOR command is not materialized to files, as when executed by the GORpipe engine. Rather, it generates RDD partitions that are provided to the Spark engine, e.g. when the results are collected through the evaluation of `snpCount`. It might help explaining this to write the `snpCount` in alternative way:

**Example 8:** Alternative form of `snpCount`

```scala
val snpCount = sgs.dataFrame("select count(*) from <(pgor [#myexons#]
  | join -segsnp -ir #dbsnp#
  | join -snpsnp -r #genes#) group by gene_symbol")
```

3 Depend on the nature of the GOR commands.
Notice, that because we are evaluating this SparkGOR expression in the context of sgs, our definitions for #myexons# and #genes# are available. The create statement may or may not have been materialized already. If not, it will be materialized when the lazy dataframe is evaluated.

As an example of how GOR can be used as tail-expression on a generic dataframe, based on myVars.parquet with variants, consider the following:

### Example 9: DataFrames with GOR tail-expression
```scala
val dbsnpDf = spark.read.parquet("dbsnp.parquet")
val myVars = dbsnpDf.gor("calc type = if(len(ref)=len(alt),'SnP','InDel')")
myVars.createOrReplaceTempView("myVars")
sgs.setDef("#VEP#","vep.gorz")
val myVarsAnno = sgs.dataFrame("select * from myVars order by chrom, pos").gor("varnorm -left ref alt | group 1 -gc ref,alt,type -set -sc rsID | rename set_rsIDs rsIDs | varjoin -r -l -e 'NA' <(gor #VEP# | select 1-alt,consequence)"")
```

Here we define the dataframe dbsnpDf from a regular Parquet file. Then we use a tail-expression to add a type column, something which is also easily done using standard map functions in conventional Spark. Next we register the dataframe as temp view and add a definition into the SparkGO session, sgs. This allows us to refer the data in SparkSQL and to order it as in GOR. Finally, we add a more complex tail-expression that left-normalizes the variants, collapses rsIDs of equivalent variants into one row, followed by a left-varjoin to annotate them with VEP-consequence. It is important to recognize that both VARJOIN and VARJOIN can only be executed on a dataframe that was generated from a SparkGOR session, because behind the scenes these commands use a genome reference build file that is defined in gorconfig.txt as shown in Ex. 7. The fact that one can indeed apply tail expressions on a regular dataframe, like dbsnpDf, is through implicit magic in Scala, however, commands that require the genome reference context will not work. Another thing worth pointing out is that when GOR generates a dataframe, the schema is automatically inferred from the query expression to generate Spark SQL row. Similarly, GOR can use row models from SparkSQL within the GOR code base, thus there is minimal "impedance mismatch".

Next we might want to persist the results to files. This can be done as show below:

### Example 10: Writing output to files
```scala
myVarsAnno.write.save("myVars.parquet")

myVarsAnno.write .format("gorz").partitionBy("chrom").save("myVars.gord")
```

The former command writes the results to a regular Parquet file. Actually, the SparkSQL engine might choose to execute myVarsAnno in parallel, hence the output myVars.parquet could be a folder of Parquet files. Similarly, we can write the output in a GOR format. We have adjusted our dictionary table format to be analogous to Parquet, i.e. myVars.gord is a folder with GORZ files as well as a GORD file as described in [7].

Our last example shows how easily GOR can now be integrated into a whole genome regression analysis workflow, written in PySpark. The workflow uses a machine learning method called REGENIE [11], which uses linear mixed models to account for population structure and relatedness, like several other similar statistical algorithms. Unlike former approaches, they build a combined phenotype predictor based on multiple local Ridge regression models, each accounting for the correlation of local linkage disequilibrium within blocks of common variants and the phenotypes. Importantly, their block based approximation approach in the model fitting step has linear computational complexity with the number of samples. Furthermore, the block based approach makes this algorithm ideally suited to take advantage of distributed computing frameworks, such as Spark. The same applies to the second step, where the adjusted phenotype model is typically used in regression against many more variants, e.g dozens of million as compared to few hundred thousand variants in the first step. A Spark version of this algorithm has been implemented in Glow and made available as PySpark code in a notebook [1]. We show here extracts of this PySpark code with focus on the parts that involve selection of the phenotypes and the setup of the variants dataframe:

### Example 11: Whole genome regression with GloWGR
```python
... import spark._jvm.org.gorpipe.spark
c = spark.sparkContext
... label_df = pd.read_csv(phenotypes_path, index_col='sample_id')
pheno_df = spark.createDataFrame(label_df)
pheno_df.createOrReplaceTempView("pheno")
sgs = SparkGOR.createSession(spark._jsparkSession)
sgs.setCreateAndDefs('
  create #pheno# = select * from pheno;
  create #samples# = nor (#pheno#);
  | inset -c sample_id varbuckets.tsv
  | select sample_id;
  create #regionsplit# = gor LDpruned_variants.gorz
  | group 1000 -count | seghist 10000"

dsl = sgs.dataFrame("gor -split [#regionsplit#] variants.gord -nf -ff [#samples#]
| varjoin -i step1_variants.gorz
| csvsel -u 3 --gc ref,alt -vs 1 varbuckets.tsv [#samples#]
| rename Chrom contigName
| rename pos start
| rename ref referenceAllele
| rename alt alternateAlleles
| `withColumn('values',chartodoublearray(col('values')))

> variant1 = _java2py(sc,dsl)
```
...variants1_dfm = variant1.withColumn('values',  
    mean_substitute(col('values')).
    filter(size(array_distinct('Values')) > 1))
...

sample_ids = sgs.dataFrame("nor [#samples]")

block_df, sample_blocks = glow.wgr.functions  
    .block_variants_and_samples(variants1_dfm,  
        sample_ids,  
        variants_per_block,  
        sample_block_count)
...

wgr_gwas = variants2_df  
    .join(adjusted_phenotypes, ['contigName'])  
    .select('contigName',  
        'start',  
        'names',  
        'label',  
        expand_struct(linear_regression_gwas(  
            variant_df.values,  
            adjusted_phenotypes.values,  
            lit[covariates.to_numpy()])))

As always, we need to import our SparkGOR library. From the Pandas phenotype structure, label_df, we create a Spark dataframe and register it as a temporary view, for it to be accessible in our SparkGOR session. As shown in the definition of #samples#, the sample IDs are found from the overlap of samples with phenotypes and those with genotypes, based on the variants bucket metadata file varbuckets.tsv. In the original notebook workflow, however, the list of sample IDs is read from the pVCF datasource header. The regions used for the parallel partitions, #regionsplit#, are defined such that there are 10k variants in each region, resulting in about 100 evenly sized partitions, if 1 million common LD-pruned variants are used for the model step. We then define our variant dataframe with GOR expression as shown with dsl. The GOR expression reads only the GORD file partitions that are needed based on the samples used, e.g. -ff [#samples#], and a simple intersect varjoin is used to extract only the variants of interest. In the GOR horizontal genotype format, the genotypes are already bi-allelic, so a multi-allelic split, as in the original notebook, is not needed.

The CSVSEL command is then used to convert multiple bucket based variant rows into a single row for each variant, with a “horizontal” values column, for the genotypes of interest and in the order specified by #samples#. In GOR, values is a regular string typed column. Therefore, we convert it to a Spark array, using the withColumn DataFrame method and chartdoubledarray, a UDF that we defined in order to format the genotype array such that it is compatible with the Glow values column. The definition of the variant input to step 2, variants2_df, is analogous to the one for step 1, except that it does not use LDpruned_variants.gorz, but rather a different file or no intersection varjoin step, to include all available variants.

Note that the GOR expression dataframes for the variants are lazy and not evaluated until the Glow regression steps fetch the variants, e.g. in the evaluation of wgr_gwas in regression step 2. Thus, there is no explicit write of intermediary temporary files here, although the execution engine may create some behind the scenes, e.g. depending on the amount of available memory for shuffle operations.

Regarding the above, there is room for optimization in the above script. The Glow method block_variants_and_samples does indeed not assume genome ordering or partition of the dataframe based on the samples, therefore it always invokes re-partitioning and ordering of the data, to split it up into block_df. In GOR, the genotype data is already stored in ordered partitions and we can easily re-partitions the subset of samples efficiently into the appropriate blocks as show below:

Example 12: Optimized block dataframes

```scala
sgs.setCreate("#samples*",  
    *nor [#samples]*
    | map -c sample_id varbuckets.tsv  
    | sort -c bucket,sample_id  
    | select sample_id*)
sgs.setCreate("#BlockRegionSplit*",  
    *gor LDpruned_variants.gorz  
    | group 1000 -count | seghist (:id)*  
    .format(variants_per_block))
sgs.setCreate("#SampleBlocks*",  
    *nor [#samples]*  
    | rownum  
    | calc sample_block div(rownum, :id)  
    | replace sample_id squote(sample_id)  
    | group -gc sample_block -lis -sc sample_id  
    | calc size listsize(lis_sample_id)  
    .format(sample_block_count))
sgs.setCreate("#RegionSampleBlocks*",  
    *nor [#BlockRegionSplit]*  
    | replace :2 #2+1 /* -p is one-based */  
    | calc header_block #1+"_"#2+"_"#3  
    | multimap -cartesian [#BlockRegionSplit#])

block_df = sgs.dataFrame(  
    parallel -parts [#RegionSampleBlocks]  
    <gor variants.gord -nf  
    -p #(!col:chrom):(col:bpStart)-#(!col:bpStop)  
    -f #(!col:ls_sample_id)  
    | varjoin -1 step1_variants.gorz  
    | csvsel -u 3 -gc ref,alt -vs 1  
    | varbuckets.tsv (nor [#samples])  
    | where sample_id in [#(!col:ls_sample_id)])  
    | calc header_block #(!col:header_block)  
    | calc sample_block #(!col:sample_block)  
    | calc size #(!col:size)  
    | rename ... as before}).withColumn(...as before)
```

Notice that we have modified the definition of #samples# such that it is ordered according the bucket structure of our variant source, variants.gord, as specified in varbuckets.tsv. This is to ensure that each GORZ bucket file is read by as few partition threads as possible. Now we define #BlockRegionSplit# to include the numbers of variants desired for each block in the model step, as specified by the variable variants_per_block. Then we group the samples into blocks in #SampleBlocks# and finally we use a Cartesian multimap-join to setup two dimensional blocks, for the variants and the samples, i.e. #RegionSampleBlocks#.

The macro command PARALLEL is then used to create as many parallel partitions as there are rows in #RegionSampleBlocks#.

---

6 SEGHIST sizes segments based on histogram equalization like approach.
allowing reference to the columns of this parts relation via 
#(col:colname) to be used as input to each query task. Notice that 
the -p option specifies the region in each block and similarly 
does the partition filtering option. -f, specify the samples that are 
needed. However, since the horizontal bucket rows store values 
from multiple samples, we extract values from the rows only 
for the samples that belong to the block. This is done with the 
OR expression, provided as the second parameter to the CSVSEL 
command. The calculation of SIZE and the two other block 
columns is to be compatible with the output from the original Glow 
method.

Finally, we write the regression results to disk, first as Parquet 
files and then as GORZ dictionary files, simply to show the 
possibilities.

**Example 13:** Writing GWAS output for many traits

```scala
wgr_gwas.createOrReplaceTempView("wgrgwas")
spark.sql("select * from wgrgwas
  order by label, chrom, pos"
).write.format("parquet").partitionBy("label")
.save("wgr_gwas.parquet")
```

```scala
gsps.dataFrame("select * from wgrgwas")
  .repartition("label")
  .gor("sort genome")
  .write.format("gorz").partitionby("label")
  .save("wgr_gwas.gord")
```

In both cases, we partition based on trait\(^7\) and order based on 
genome position. In the latter case, we show an example where 
a GOR tail-expression is used on a dataframe, to do the ordering 
within each trait. In supplementary material \([5]\), we show more 
examples including how GWAS association results, from close 
to 1500 PheCode traits defined from the UKBB data, can be 
interrogated using the query capabilities of SparkGOR.

On small datasets when there are few samples and variants in the 
model step, the speed difference between the approach shown in 
Ex. 11 and Ex. 12 may not play a major role and in general, the 
steps involving the RidgeReducer()\(^8\) may overshadow the shuffling 
overhead in ordering the variants, especially when multiple trait 
are analyzed together. Nevertheless, the above example presents a 
relatively easy path to integrate GOR in an effective manner into 
a Glow based algorithm. Clearly, the SparkGOR SDK integration 
opens up multiple options and choices for working with genome 
data using a growing number of libraries that are built for the Spark 
platform, such as Glow, ADAM, GATK, and Hail.

4 DISCUSSION

In this paper, we have presented an integration of GOR with 
Spark, first as a unified relational query engine, mixing declarative 
SparkSQL and GORpipe queries, and secondly as a library to 
work with Spark dataframes, e.g. in Scala or Python. We have 
discussed the similarities between these two architectures as well 
as their fundamental differences, such as their approach for joining 
relations. It is our belief that each approach brings its own strengths 
and weaknesses, all depending on the use-cases at hand. This work 
was however not intended to be a formal comparison or benchmark of 
the two architectures.

Overall, we can however say that many use-cases based on 
range joins and range access are more efficient with GOR and 
less dependent on memory caching. For instance, the dynamically 
optimized seek-scan approach in GORpipe makes is possible join 
the entire ClinVar database, with few hundred thousand unevenly 
distributed variants, against a table with seven hundred million 
annotated variants, in about five seconds on a MacBook Pro. This 
takes ten times longer time in SparkSQL using the same hardware, 
even when using its most effective broadcast join approach. 
Likewise, queries that make fuzzy join on genome segments, such as 
exons or genes, or use a single range scan invoked from a genome 
scanner, run very fast in GOR but are significantly less efficient 
in Spark. Hence, de-normalization is often needed in SparkSQL 
to avoid genome range joins, thus relying on columnar access and 
efficient filtering.

Filtering speed of rows is significantly faster in SparkSQL, 
especially if the data has been cached to memory or if the filter is 
highly selective and can take advantage of row groups metadata in 
the Parquet format. Nevertheless, in the latter case, the performance 
ends up being sensitive to the ordering strategy of the table, as 
highlighted in some of our examples. Indeed, the partition strategy 
does play a major role for both systems, especially for large data 
sets. Compression ratio in GORZ and Parquet is similar while the 
computation cost of writing is less in GORZ. Also, Spark demands a 
significant amount of memory in order to work well and the Parquet 
driver seems to have a significantly higher memory footprint per file 
than the GORZ driver.

There have been efforts to address some of the above 
issues. For instance, the ADAM project \([10]\) has introduced a 
genome range partition scheme extension to Parquet for their 
GenomicDataset, allowing for a more efficient filtering by 
overlapping regions. In particular, they report that the time of range 
scans goes from few minutes to few seconds. For comparison, a 
typical range scan executed from a GOR based genome browser, 
fetching 1000 rows from a Clinvar GORZ table takes less than 
100msec.

Their approach is indeed similar to our GOR dictionary files, 
i.e. the output of PGOR queries where the file partitions and 
corresponding genome range is stored in a GORD metadata file. 
We have also started to experiment with dictionaries based on Parquet 
files (currently coined GORP), in order to non-intrusively leverage 
Parquet columnar projection and predicate push-down within GOR. 
However, since the GOR query logic is based on seek-able ordered 
iterators\(^9\), it must be done without introducing longtime blockage from 
iterators that use the Parquet filtering mechanism. By using 
genome ordered and moderately sized range partitions and by 
extending our iterator interface, such that iterators provide position 
progress information to its consumer, we have been able to adopt the 
Parquet driver with our execution patterns, i.e. where data is merged 
from multiple files and dynamic seek-scan decisions are made based 
on the streaming progress. Similar blockage issues come up in 
relation to merge patterns from federated ordered iterators, that 
implement predicate push-downs or more complex query logic such

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\(^7\) Here called label to be compatible with the original notebook.

\(^8\) A step not shown here but in the original GloWGR notebook.

\(^9\) Different from the standard Volcano iterator model\([6]\)
as grouping, patterns that can be of value to increase parallelism or reduce the impact of network bottlenecks[18].
A detailed description of the above development is outside the scope of this work and awaits publication elsewhere. Same applies to other areas of further integration that lie ahead, for instance in relation to increased usage and generation of metadata to automate GOR execution within the Spark engine. It is our hope that open-sourcing SparkGOR will spur innovation and widen the interest in the unified platform.

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