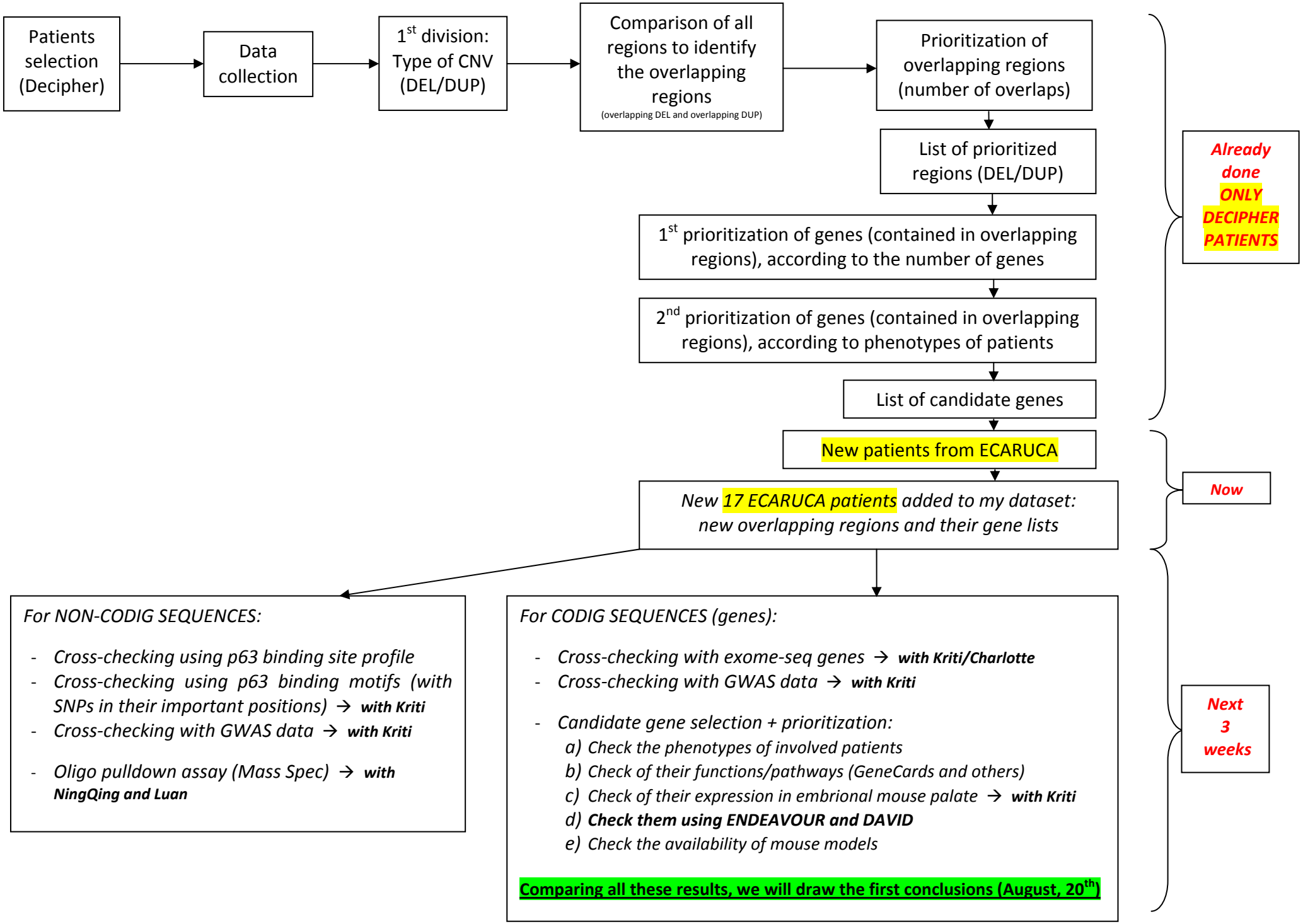
Patients selection (Decipher)
Data collection
1st division: Type of CNV (DEL/DUP)
Comparison of all regions to identify the overlapping regions
(overlapping DEL and overlapping DUP)
Prioritization of overlapping regions (number of overlaps)
List of prioritized regions (DEL/DUP)
1st prioritization of genes (contained in overlapping regions), according to the number of genes
2nd prioritization of genes (contained in overlapping regions), according to phenotypes of patients
List of candidate genes
Already done ONLY DECIPHER PATIENTS
New patients from ECARUCA
New 17 ECARUCA patients added to my dataset: new overlapping regions and their gene lists
Now
For NON-CODIG SEQUENCES:
- Cross-checking using p63 binding site profile
- Cross-checking using p63 binding motifs (with SNPs in their important positions) → with Kriti
- Cross-checking with GWAS data → with Kriti
- Oligo pulldown assay (Mass Spec) → with NingQing and Luan
For CODIG SEQUENCES (genes):
- Cross-checking with exome-seq genes → with Kriti/Charlotte
- Cross-checking with GWAS data → with Kriti
- Candidate gene selection + prioritization:
a) Check the phenotypes of involved patients
b) Check of their functions/pathways (GeneCards and others)
c) Check of their expression in embrional mouse palate → with Kriti
d) Check them using ENDEAVOUR and DAVID
e) Check the availability of mouse models
Comparing all these results, we will draw the first conclusions (August, 20th)
Next 3 weeks