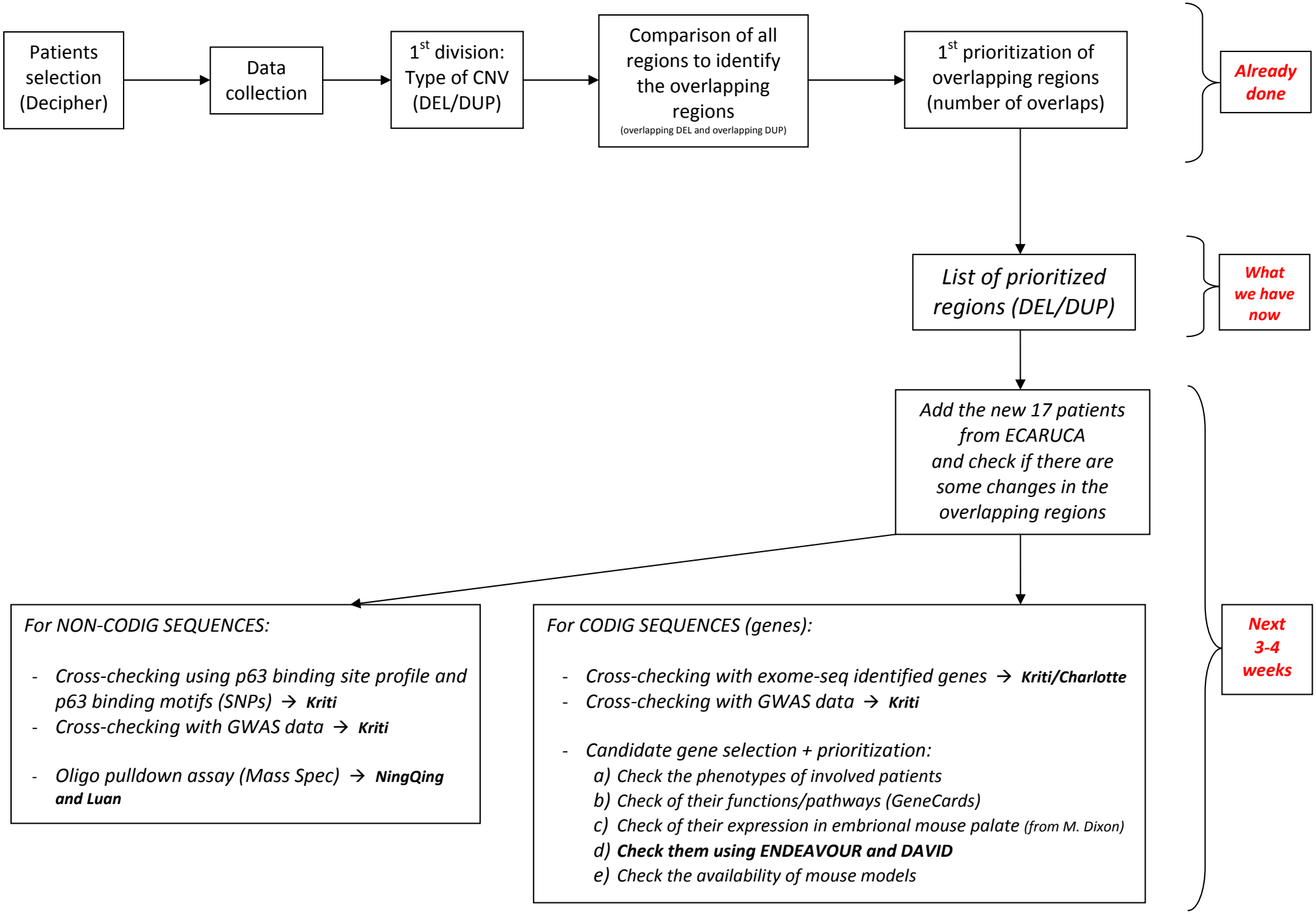
Patients selection (Decipher)
Data collection
1st division: Type of CNV (DEL/DUP)
Comparison of all regions to identify the overlapping regions
(overlapping DEL and overlapping DUP)
1st prioritization of overlapping regions (number of overlaps)
Already done
List of prioritized regions (DEL/DUP)
What we have now
Add the new 17 patients from ECARUCA and check if there are some changes in the overlapping regions
Next 3-4 weeks
For NON-CODIG SEQUENCES:
- Cross-checking using p63 binding site profile and p63 binding motifs (SNPs) → Kriti
- Cross-checking with GWAS data → Kriti
- Oligo pulldown assay (Mass Spec) → NingQing and Luan
For CODIG SEQUENCES (genes):
- Cross-checking with exome-seq identified genes → Kriti/Charlotte
- Cross-checking with GWAS data → Kriti
- Candidate gene selection + prioritization:
a) Check the phenotypes of involved patients
b) Check of their functions/pathways (GeneCards)
c) Check of their expression in embrional mouse palate (from M. Dixon)
d) Check them using ENDEAVOUR and DAVID
e) Check the availability of mouse models