*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***

Patients selection

(Decipher)

Data collection

1st division:

Type of CNV

(DEL/DUP)

***Already done***

Comparison of all regions to identify the overlapping regions

(overlapping DEL and overlapping DUP)

*For CODIG SEQUENCES (genes):*

* *Cross-checking with exome-seq identified genes* 🡪 ***Kriti/Charlotte***
* *Cross-checking with GWAS data* 🡪 ***Kriti***
* *Candidate gene selection + prioritization:*

1. *Check the phenotypes of involved patients*
2. *Check of their functions/pathways (GeneCards)*
3. *Check of their expression in embrional mouse palate (from M. Dixon)*
4. ***Check them using ENDEAVOUR and DAVID***
5. *Check the availability of mouse models*

***What we have now***

***Next***

***3-4 weeks***

*Add the new 17 patients from ECARUCA*

*and check if there are some changes in the overlapping regions*

*List of prioritized regions (DEL/DUP)*

1st prioritization of overlapping regions

(number of overlaps)