Galactosemia Testing Algorithm

- Clinical suspicion of classic galactosemia
- Positive newborn screen for galactosemia
- Investigation of possible carrier status

Order GCT / Galactosemia Reflex, Blood

Galactose-1-phosphate uridylyltransferase (GALT) enzyme analysis performed

GALT <24.5 nmol/h/mg of hemoglobin
- GALT gene analysis (14-mutation panel) is automatically performed
  - Possible causes:
    - Classic galactosemia
    - Duarte variant galactosemia
    - Carrier:
      - Classic galactosemia mutation
      - Duarte mutation
  - Concordant enzyme and mutation results
    - Diagnostic of disease or carrier status
  - Discordanent enzyme and mutation results
    - 2 mutations identified
      - Diagnostic of disease
    - 1 mutation identified and concordant enzyme results
      - Carrier status confirmed
    - 0–1 mutations identified and discordant enzyme results
      - To resolve discrepant enzyme and mutation results, consider GALTP / Galactose-1-Phosphate Uridyltransferase Biochemical Phenotyping, Erythrocytes

GALT ≥24.5 nmol/h/mg of hemoglobin
- Classic galactosemia excluded
  - Was testing ordered to follow-up an abnormal newborn screen?
    - NO
      - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
      - Normal
        - GALP normal
          - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
          - Elevated
            - GALP elevated
              - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
              - Normal
                - GALP normal
                  - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                  - Elevated
                    - GALP elevated
                      - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                      - Normal
                        - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                        - Elevated
                          - GALP elevated
                            - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                            - Normal
                              - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                              - Elevated
                                - GALP elevated
                                  - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                  - Normal
                                    - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                    - Elevated
                                      - GALP elevated
                                        - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                        - Normal
                                          - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                          - Elevated
                                            - GALP elevated
                                              - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                              - Normal
                                                - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                - Elevated
                                                  - GALP elevated
                                                    - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                    - Normal
                                                      - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                      - Elevated
                                                        - GALP elevated
                                                          - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                          - Normal
                                                            - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                            - Elevated
                                                              - GALP elevated
                                                                - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                - Normal
                                                                  - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                  - Elevated
                                                                    - GALP elevated
                                                                      - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                      - Normal
                                                                        - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                        - Elevated
                                                                          - GALP elevated
                                                                            - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                            - Normal
                                                                              - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                              - Elevated
                                                                                - GALP elevated
                                                                                  - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                  - Normal
                                                                                    - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                    - Elevated
                                                                                      - GALP elevated
                                                                                        - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                        - Normal
                                                                                          - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                          - Elevated
                                                                                              - GALP elevated
                                                                                               - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                               - Normal
                                                                                                 - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                 - Elevated
                                                                                                     - GALP elevated
                                                                                                         - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                         - Normal
                                                                                                           - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                           - Elevated
                                                                                                               - GALP elevated
                                                                                                                 - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                                 - Normal
                                                                                                                   - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                                   - Elevated
                                                                                                                       - GALP elevated
                                                                                                                            - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                                            - Normal
                                                                                                                                - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                                                - Elevated
                                                                                                                                    - GALP elevated
                                                                                                                                        - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                                                        - Normal
                                                                                                                                            - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                                                            - Elevated
                                                                                                                                                - GALP elevated
                                                                                                                                                    - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                                                                    - Normal
                                                                                                                                                           - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                                                                           - Elevated
                                                                                                                                                               - GALP elevated
                                                                                                                                                                - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                                                                                - Normal
                                                                                                                                                                         - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                                                                                         - Elevated
                                                                                                                                                                             - GALP elevated
                                                                                                                                                                - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                                                                                                - Normal
                                                                                                                                                                                 - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                                                                                                                                                                                 - Elevated
                                                                                                                                                                                     - GALP elevated
                                                                                                                                                    - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                                                                                    - Normal
                          - Rule-out galactokinase (GALK) deficiency by ordering GALP / Galactose-1-Phosphate (Gal-1-P), Erythrocytes
                          - Elevated
                            - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                              - Normal
                                - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency
                              - Elevated
                                - Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency

- * The detection rate of this gene analysis is ~80%-90% for classic galactosemia mutations.