Test Retirement: Frequently Asked Questions

1. **What tests are being retired?**

   Cytogenetics:

   Rapid Aneuploidy FISH (3582)

   Amniotic Fluid AFP (2122)

   AChE (automatic reflex after AF-AFP) (1952-1)

   Chromosome Analysis, Amniotic Fluid (2136)

   Chromosome Analysis, CVS (A587)

   Chromosome Analysis, Products of Conception (1053)

   Chromosome Analysis, Peripheral Blood (0559)

   Rule out Mosaicism, Chromosome Analysis, Blood (T982)

   Send-out Prenatal (used for infectious studies) (728)

   Xpanded/Repro/Insights:

   Xpanded HereditaryCancer – Singleton (TH42a)

   Xpanded HereditaryCancer – Trio (TH42b)

   Xpanded HereditaryCancer following GeneDx HereditaryCancer Panel–Singleton (TH42d)

   Xpanded HereditaryCancer following GeneDx Hereditary Cancer Panel – Trio (TH42e)

   ReproXpanded – Individual (J776)

   ReproXpanded – Couple (J842)

   XomeDx®Insights (TH90)

2. **Why is GeneDx retiring these tests?**

   Aneuploidy FISH, Amniotic fluid AFP, AChE, and chromosome analysis are not performed at GeneDx. These tests were previously sent to a reference laboratory. We've ended our relationship with the reference laboratory so are retiring these test codes.

3. **Is there another test I can order in place of the one being retired?**

   The American College of Obstetricians and Gynecologists (ACOG) has recommendations around prenatal chromosomal microarray (CMA) and karyotype that provide some helpful context.

   ACOG:

- Prenatal chromosomal microarray analysis is recommended for a patient when a fetus has one or more major structural abnormalities identified on ultrasonographic examination and who is undergoing invasive prenatal diagnosis. This test typically can replace the need for fetal karyotype.
- In a patient with a structurally normal fetus who is undergoing invasive prenatal diagnostic testing, either fetal karyotyping or a chromosomal microarray analysis can be performed.
- Chromosomal microarray analysis of fetal tissue (i.e., amniotic fluid, placenta, or products of conception) is recommended in the evaluation of intrauterine fetal death or stillbirth when further cytogenetic analysis is desired because of the test's increased likelihood of obtaining results and improved detection of causative abnormalities.

Helpful Links:

- https://journals.lww.com/greenjournal/Fulltext/2016/12000/Committee_Opinion_No__682_Summary__Microarrays_and.52.aspx
- ACMG: https://www.gimjournal.org/article/S1098-3600(21)02687-3/fulltext

1. Rapid Aneuploidy FISH (3582)

Replacement: no rapid replacement, however, prenatal chromosomal microarray (460 or 410) provides information on aneuploidy or other copy number variants

2. Amniotic Fluid AFP (2122)

No replacement

3. AChE (automatic reflex after AF-AFP) (1952-1)

No replacement

Chromosome Analysis, Amniotic Fluid (2136)

Replacement: Prenatal chromosomal microarray (460 or 410). Microarray is a superior test for detecting copy number imbalances. It is not able to detect balanced structural rearrangements (translocations, inversions). FISH follow-up testing is available to interrogate for known familial rearrangements.

Chromosome Analysis, CVS (A587)

Replacement: Prenatal chromosomal microarray (460 or 410). Microarray is a superior test for detecting copy number imbalances. It is not able to detect

balanced structural rearrangements (translocations, inversions). FISH follow-up testing is available to interrogate for known familial rearrangements.
Chromosome Analysis, Products of Conception (1053) Replacement: Prenatal chromosomal array (460). We will accept products of conception (POC) for chromosomal microarray. Microarray is a superior test for detecting copy number imbalances. It is not able to detect balanced structural rearrangements (translocations, inversions). FISH follow-up testing is available to interrogate for known familial rearrangements.
Chromosome Analysis, Peripheral Blood (0559) Replacement: Chromosomal microarray (910)
Rule out Mosaicism, Chromosome Analysis, Blood (T982) No replacement. Microarray is not ideal for low level mosaicism.
Send-out Prenatal (used for infectious studies) (728) No replacement.

4. **What alternative labs can be utilized if there are no replacement tests being offered?**

   FISH and Chromosomes are available at various commercial laboratories including Quest, LabCorp/Integrated Genetics, BioReference.

5. **When is the last day I can order these tests?**

   The last day to accept samples is March 31, 2023. If your paper order form accompanies a sample and is received by March 31, 2023, GeneDx will accept the order. Any order received after this date will be cancelled and samples returned.

6. **What is the last day to accept samples?**

   March 31, 2023

7. **When can I expect my report?**

All reports will be sent within regular turnaround times specific to test category, if samples were received by 3/31/23.

8. **Will GeneDx be accepting redraws after the sample cutoff date of 3/31/23?**

   No

9. **When will FISH testing be offered?**

   GeneDx will only offer FISH testing when it is a follow up to a GeneDx molecular test.

10. **Will prenatal targeted testing still be offered to pregnancies at-risk to inherit a familial variant?**

    GeneDx will still offer targeted testing for familial variants. Acceptable sample types include direct amniotic fluid, chorionic villi, extracted DNA or cultured cells. Please contact the prenatal GC team (prenatal@genedx.com) for any questions about prenatal targeted testing.

    GeneDx will also continue to offer all other molecular testing that is currently on our prenatal testing menu.

11. **I do not want to get set up with a different Lab. Will GeneDx send samples to a different Lab on my behalf?**

    Unfortunately, we are unable to send samples to another lab for the tests we are retiring. GeneDx will not split samples, so we are not able to accommodate requests to perform molecular testing at GeneDx and send a portion of the sample to an external laboratory for other tests.

12. **Are specific sample types no longer being accepted?**

    GeneDx will continue to accept prenatal sample types for molecular tests on the testing menu, including direct amniotic fluid or CVS samples, cultured cells, and extracted DNA.

    It is recommended that amniotic fluid samples with visible blood not be sent, as these direct samples are expected to have significant maternal cell contamination precluding the ability to perform fetal testing.

13. **What should I do if I want to order FISH and/or chromosomes in addition to molecular testing at GeneDx?**

    GeneDx is no longer able to perform testing or coordinate sendouts to a referral lab for FISH, chromosome analysis, or other tests that are being retired.

    If these tests are being performed at an outside laboratory, the amnio or CVS samples should be sent to a laboratory who can prepare cultured flasks or DNA to send to GeneDx for the molecular testing.

14. **What sample type should I send if I am ordering a fetal/prenatal exome?**
    Direct amniotic fluid or chorionic villi, cultured flasks of chorionic villi or amniotic fluid, extracted DNA, and products of conception (POC) are all acceptable sample types.

15. What if I have additional questions?

For additional questions please call or email us:

Phone: **(301) 519-2100**, option 3
Toll Free: **(888) 729-1206**, option 3
Email: **support@genedx.com**
Monday to Friday 8 am - 8 pm Eastern