Cell Free DNA (cfDNA)

Why in news?

Cell free DNA is found to be promising in the field of disease discovery.

What is cfDNA?

> *DNA is a molecule that carries genetic information for the development and functioning of an organism.*

- Cell-free DNA (cfDNA) refers to small DNA fragments found in the bloodstream and other bodily fluids, such as spinal fluid and urine.
- These fragments are derived from various sources, including the breakdown of cells, the release of DNA from damaged or dying cells, and the shedding of DNA by normal cells.
- The presence of cfDNA in bodily fluids is an indication of various processes that include physical injury, inflammation, and cancer.

What are the applications of cfDNA?

- **Non-invasive prenatal test (NIPT) -** NIPT is a screening test that can be performed during pregnancy to assess the risk of chromosomal abnormalities in the fetus, such as
  - Down syndrome (trisomy 21)
  - Edwards syndrome (trisomy 18)
  - Patau syndrome (trisomy 13)
- **Cancer treatment-** The presence of cfDNA with specific genetic abnormalities can indicate the presence of cancer.
- The analysis of cfDNA can be used to diagnose cancer, monitor the treatment effectiveness, and detect recurrence after treatment.
  - **GEMINI-** Genome-wide Mutational Incidence for Non-Invasive detection of cancer adopts a whole-genome-sequencing approach to cfDNA extracted from patients.

- **Biomarker-** CfDNA finds its application in the neurological disorders like Alzheimer's disease, neuronal tumours, stroke, traumatic brain injury, and even metabolic disorders like type-2 diabetes and non-alcoholic fatty liver disease.
- **Heart attack**- The analysis of cfDNA has been studied as a potential tool for the

diagnosis and prognosis of acute myocardial infarction (heart attack).

- **Stroke**- The presence of cfDNA in the blood is an indicator of brain injury in stroke patients, and the analysis of cfDNA has been studied as a potential tool for the diagnosis and prognosis of stroke.
- **Autoimmune diseases**- The analysis of cfDNA is a potential tool for diagnosing and monitoring autoimmune diseases, such as rheumatoid arthritis and systemic lupus erythematosus.
- **Pregnancy complications**- The analysis of cfDNA is a potential tool for diagnosing and monitoring complications during pregnancy, such as preterm labor and preeclampsia.
- **Organ transplantation-** It helps in understanding why a body is rejecting a transplanted organ.
- DNA obtained from the donor who is donating the organ – called donor-derived cfDNA, dd-cfDNA – could provide an early yet accurate estimate of how well the organ is being taken up.

Circulating tumor DNA (ctDNA)

- ctDNA refers to a small subset of the cfDNA and consists of small DNA fragments released into the bloodstream by cancer cells.
- ctDNA can be detected in the blood of individuals with cancer and can provide information about the characteristics of the cancer.
- **Size-** ctDNA fragments are typically smaller than normal DNA fragments due to the presence of breaks and mutations in the DNA.
- **Composition-** ctDNA comprises both normal DNA and DNA with mutations or abnormalities.
- **Stability-** ctDNA is relatively stable in the bloodstream and can be detected for an extended period after it is released from the cancer cells.
- **Concentration-** The concentration of ctDNA in the bloodstream can vary depending on the stage and size of the cancer.
- **Heterogeneity**- ctDNA can be highly heterogeneous, meaning it can contain various mutations and abnormalities making it challenging to detect and analyze accurately.

Reference

[The Hindu- cfDNA cancer screening]

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