Somatic Genetic Variants

For Prelims: Genome sequencing, **Cancer,** Somatic Genetic Variants, Germline cells, DNA replication.

For Mains: Harnessing Somatic Genetic Variants for Human Health Advancement

Source: TH

Why in News?

Recent advances in **genome sequencing** unveil the impact of **somatic genetic variants on** human health**, from** cancer **development to immune disorders**, driving innovation in disease detection and treatment strategies.

What are Somatic Genetic Variants?

- **About:**
  - Somatic genetic variants also known as somatic mutations **refer to alterations in the DNA sequence that occur specifically within the cells** of an individual's body (somatic cells), **excluding the** germline cells **(sperm and egg cells).**
  - **Somatic genetic mutations occur after birth during development and are not inherited from parents.**
- **Somatic Mutation Progression:**
  - The **human genome consists of 23 pairs of** chromosomes, inherited from each parent, forming the **blueprint of our genetic identity.**
    - Following the fertilization of an egg cell by a sperm cell, the resultant single cell amalgamates genetic material from both parents.
    - Through subsequent rounds of division, **this initial cell proliferates extensively,** ultimately generating the **countless trillions of cells constituting the human body.**
  - During the **process of** DNA replication**,** the incorporation of errors is notably minimized by error-correcting proteins. Nevertheless, a minute error rate persists and occurs at different times, **contributing to the emergence of somatic genetic mutations.**
    - As cells continue to renew and replace old ones throughout life, errors keep occurring, leading to the **gradual accumulation of somatic mutations over time.**
    - This is why **differences in genetic makeup are observed between different tissues in the body as people get older.**
- **Influence of Somatic Genetic Variants on Human Health:**
  - **Cancer Development:** Somatic genetic variants can drive the **uncontrolled cell growth and division characteristic of cancer,** leading to **tumor** formation.
  - **Neurological Disorders**: Accumulated somatic mutations in **brain cells can contribute to neurological conditions,** affecting cognitive and motor functions.
  - **Aging and Tissue Function:** Gradual accumulation of somatic mutations with age can **impair tissue function** and contribute to age-related diseases.

        - **Immune System Dysfunction**: Somatic variants can **disrupt immune cell development and function**, leading to autoimmune disorders and **immunodeficiencies.**
- **Harnessing Somatic Genetic Variants for Human Health Advancement:**
    - **Disease Biomarkers:** Somatic variants can serve as **diagnostic and prognostic markers for diseases.**
        - Detecting specific mutations can aid in **early disease detection** and predicting disease progression.
    - **Precision Medicine:** Knowledge of an individual's somatic mutations allows **personalized treatment plans.**
        - Tailoring therapies to a patient's unique genetic makeup can **enhance treatment outcomes.**
    - **Aging and Longevity:** Studying somatic mutations associated with aging can shed light on the **aging process and age-related diseases**, potentially leading to interventions for healthier aging.
    - **Solution of Genetic Disease:** In some instances, somatic mutation brings a deleterious change to a normal one, a phenomenon known as **revertant mosaicism**.
        - E.g., **Around 10% of cases of Wiskott-Aldrich syndrome**, a rare genetic immunodeficiency, have been found to have revertant mosaicism, as a result alleviating the severity of the disease in many individuals.

UPSC Civil Services Examination, Previous Year Question (PYQ)

Q.1 With reference to agriculture in India, how can the technique of 'genome sequencing', often seen in the news, be used in the immediate future? (2017)

1. Genome sequencing can be used to identify genetic markers for disease resistance and drought tolerance in various crop plants.
2. This technique helps in reducing the time required to develop new varieties of crop plants.
3. It can be used to decipher the host-pathogen relationships in crops.

Select the correct answer using the code given below:

(a) 1 only
(b) 2 and 3 only
(c) 1 and 3 only
(d) 1, 2 and 3

Ans: (d)

Q2. Consider the following statements: (2022)

1. DNA Barcoding can be a tool to:
2. Assess the age of a plant or animal.
3. Distinguish among species that look alike.
4. Identify undesirable animal or plant materials in processed foods.

Which of the statements given above is/are correct?

(a) 1 only
(b) 3 only
(c) 1 and 2
(d) 2 and 3

Ans: (d)