

1. Prenatal Diagnosis:

Definition: A range of procedures used to detect genetic abnormalities and structural anomalies early in an embryo or fetus.

Benefits Include:

- ✓ Timely Intervention: Allows for early treatment or management before or after birth.
- ✓ Informed Decisions: Helps parents decide on the continuation of a pregnancy if a condition is diagnosed.

Indications for Prenatal Diagnosis:

Risk Factors:

- a. Advanced maternal age
- b. Previous child with a chromosomal abnormality
- c. Multiple pregnancies (twins or more)
- d. Family history of single gene disorders, neural tube defects, or other congenital abnormalities
- e. History of miscarriages
- f. Additional high-risk factors such as consanguinity or maternal illnesses

Methods Employed:

A. Invasive Techniques:

1. Amniocentesis: Extraction of amniotic fluid to test fetal cells.
2. Cordocentesis: Sampling of fetal blood from the umbilical cord.
3. Chorionic Villus Sampling (CVS): Collection of placental tissue.
4. Fetal Tissue Biopsy: Direct sampling of fetal cells.
5. Coelocentesis: Rarely used method for fetal fluid extraction.

B. Non-Invasive Techniques:

1. Ultrasonography: Imaging to check for structural anomalies.
2. MRI: Detailed imaging when needed.
3. Cell-Free Fetal DNA: Analysis of fetal DNA circulating in the mother's blood.
4. Triple Test: Maternal blood test that measures specific markers.

2. Stem Cell Technology:

Definition & Characteristics

Stem Cells: Cells with the unique ability to self-renew (divide continuously) and differentiate into various specialized cell types.

Types of Stem cells based on Potency

1. Totipotent stem cells : Each cell in the 8-cell embryo can generate every cell in the embryo as well as the extra-embryonic tissues
2. Pluripotent stem cells : The embryonic stem cells which can generate every cell in the body except extra-embryonic tissues .
3. Multipotent stem cells : tissue-specific stem cells (Adult)
4. Unipotent stem cells : only generate one cell type

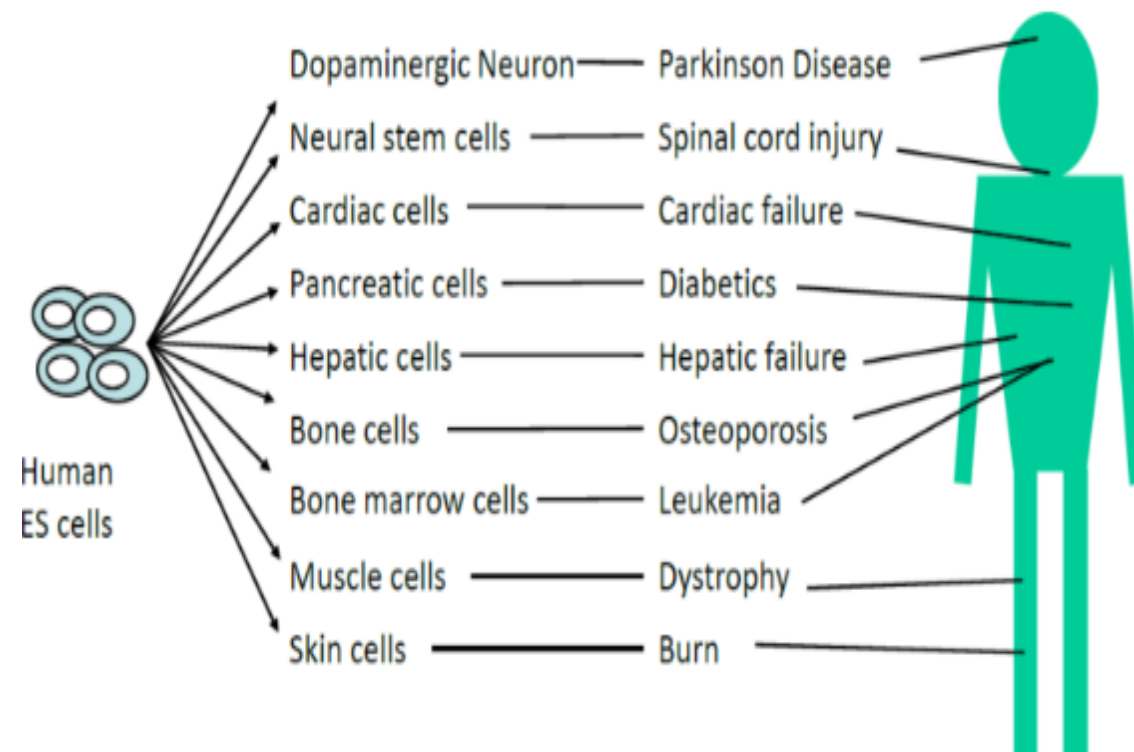
Traits of stem cells :

1. Self-renewal: They can replicate for extended periods.
2. Unspecialized Nature: They are not yet committed to a specific function, allowing them to become specialized cells as needed.
3. Differentiation Potential: They can generate a wide range of specialized tissues.

Applications in Medicine:

1. Cancer Treatment (Leukemia): Stem cells (from bone marrow transplants) are used to replace diseased blood cells in leukemia patients.
2. Heart Disease: Adult bone marrow cells injected into the heart show potential in improving heart function after failure or heart attack.
3. Parkinson's Disease: Treatment strategies focus on replacing or repairing dopamine-producing neurons affected in Parkinson's.
4. Rheumatoid Arthritis: Stem cells may help repair eroded cartilage in joints affected by this autoimmune condition.

Regenerative Medicine & ES Cell

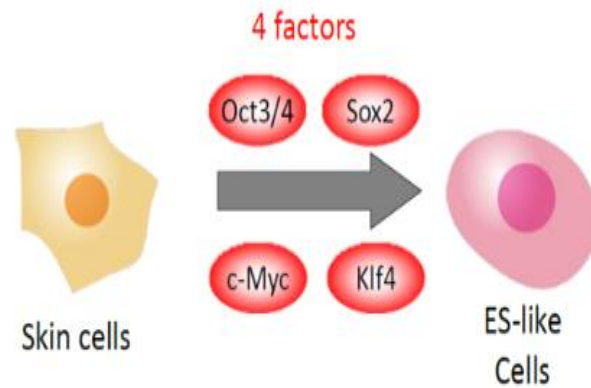


Advancements & Ethical Considerations:

1. Advancements in Stem Cell Research

Induced Pluripotent Stem Cells (iPSCs): Created by reprogramming ordinary fibroblast cells using specific DNA factors. Function like embryonic stem cells but without the ethical concerns of embryo destruction.

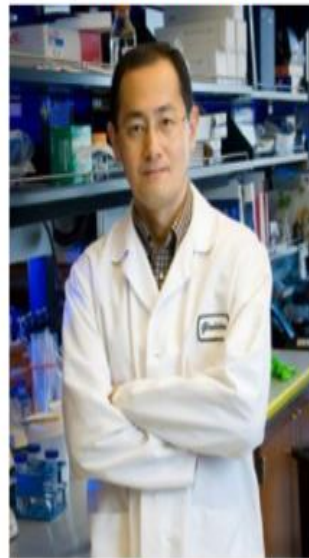
Induced Pluripotent Stem (iPS) Cells



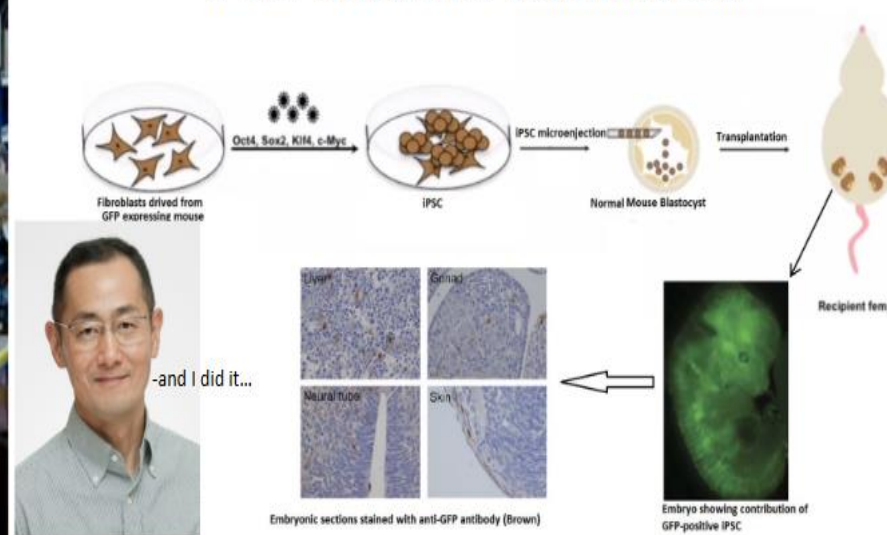
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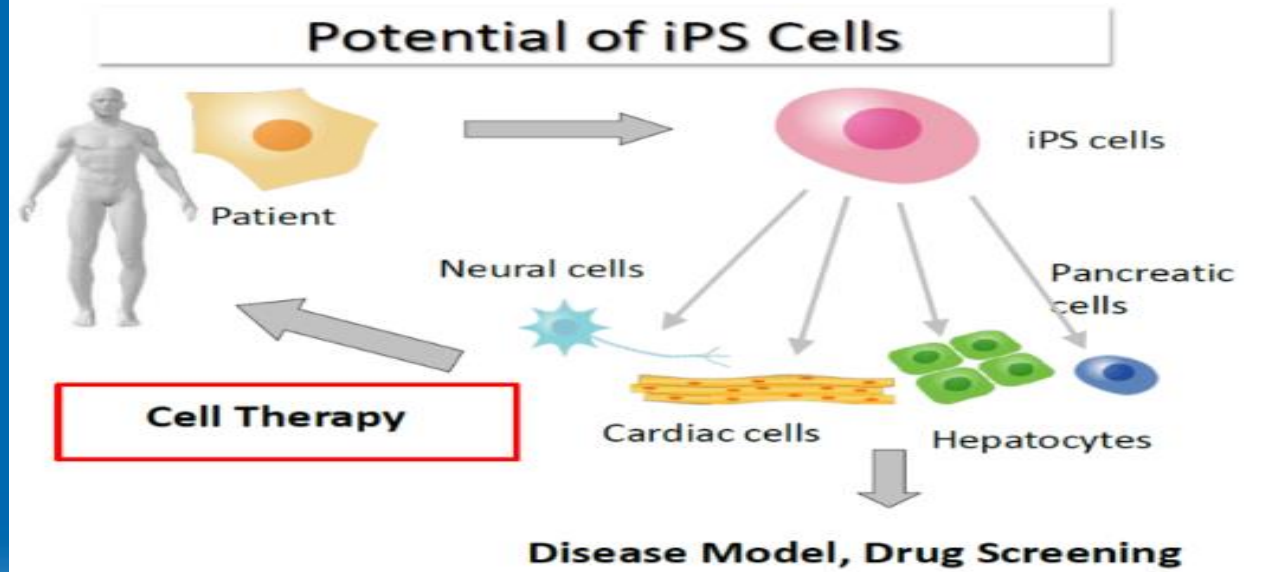
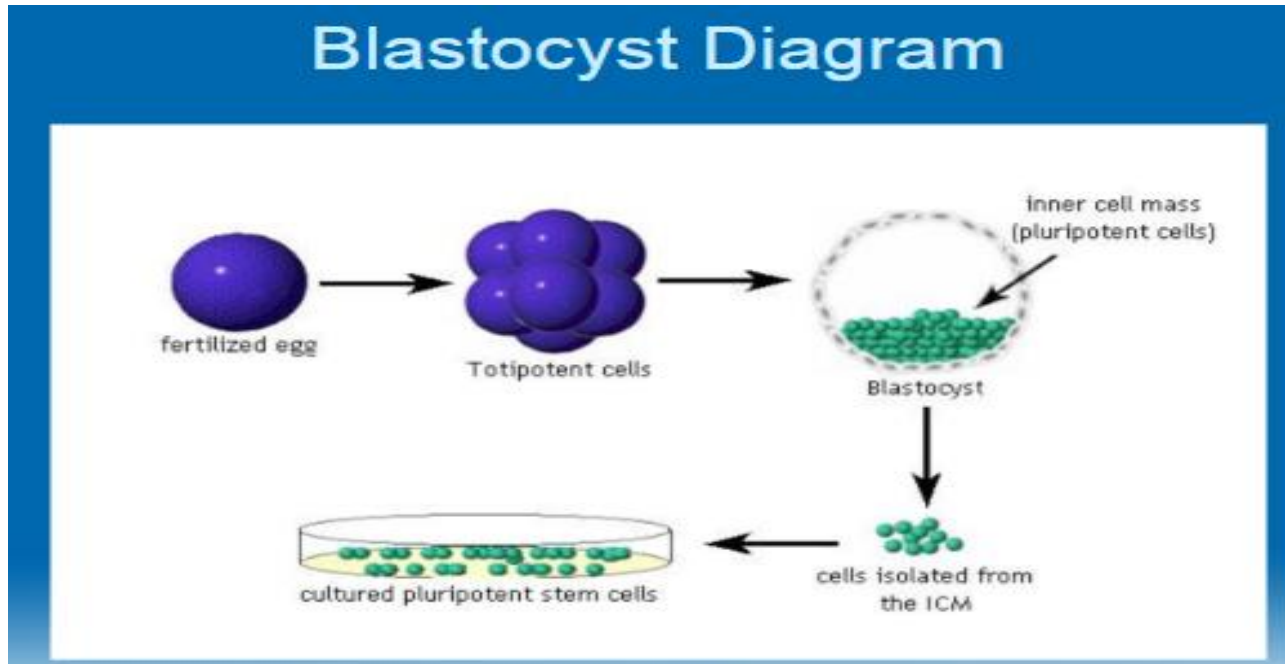
Human 2007

Dr. Shinya Yamanaka



iPSC Functional confirmation





2. Ethical Debates

Controversial Sources:

Embryonic Stem Cells (ESCs): Ethical concerns due to the destruction of human embryos.

Embryonic Germ Cells: Ethical acceptability depends on whether the source is elective or spontaneous abortion.

Widely Accepted Sources: Stem cells derived from umbilical cord blood, placental tissues, and adult tissues are generally considered ethical.

1. Gene therapy

Gene therapy involves inserting, altering, or removing genes within an individual's cells to correct defects that cause disease. Its main aim is to replace or repair mutated genes with functional ones.

Approaches:

- Insertion of Functional Genes: The most common method where a new gene is introduced to replace a mutated one.
- Direct Correction or Modification: Some techniques correct the mutation directly or modify normal genes to counteract viral infections.

Types of Gene Therapy:

- Germ Line Gene Therapy: Functional genes are introduced into sperm or egg cells. The genetic modifications become heritable, passing on to subsequent generations.
- Somatic Gene Therapy: Therapeutic genes are delivered into non-reproductive (somatic) cells. Effects are limited to the treated individual without inheritance by offspring.

Delivery Methods (Vectors):

A. Viral Methods: Use viruses (e.g., retroviruses, adenoviruses, herpes simplex virus) that naturally infect cells to deliver the therapeutic gene. Viruses can follow lytic cycles (rapid replication and cell lysis) or lysogenic cycles (integrating into the host genome for a prolonged period)

B. Non-Viral Methods:

- Physical Methods: Such as electroporation, gene gun, sonoporation, and magnetofection to introduce naked DNA into cells.
- Chemical Methods: Utilize compounds like oligonucleotides, lipoplexes, polyplexes, and dendrimers.
- Hybrid Methods: Combine techniques (e.g., virosomes that mix liposomes with inactivated viruses) to improve gene transfer efficiency.

Challenges:

- Short-lived Effect: The therapeutic DNA may not remain functional long-term, often necessitating repeated treatments.
- Immune Response: The body's defense system can attack the introduced gene material or viral vectors, reducing effectiveness.
- Viral Vector Risks: Potential toxicity, immune reactions, and the risk of the vector reverting to a disease-causing form.
- Complexity of Multigene Disorders: Conditions caused by multiple gene variations (e.g., heart disease, diabetes) are more challenging to treat.
- Insertional Mutagenesis: Improper integration of the therapeutic DNA can disrupt normal genes (such as tumor suppressor genes), potentially leading to cancer.