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# Gene Therapy for "Butterfly Children"

Videos of the "butterfly children" are difficult to watch. The name comes from the delicate skin of people who have **epidermolysis bullosa**, which is reminiscent of the fragility of a butterfly's wings. The slightest touch causes painful blisters and peeling skin. Parents can be carriers of **Recessive dystrophic epidermolysis bullosa (RDEB)** without having any of the effects. RDEB affects about 1 in every 20,000 births in the United States.

Parents of RDEB children provide intensive care for their children, which includes daily full body bandage changes and use of antibiotics and antiseptics. These bandages are made of special polymers that have low adhesion so they don't tear the skin if a parent pulls a bandage off.

The daily bandage changes, pricking of blisters, and removing flaky skin aren't all the requirements. Bathing can take hours. Complications include anemia, malnutrition from mouth and esophagus sores that make eating nearly impossible, and skin cancer. Scarring of skin can cause the fingers and toes to fuse together creating "mitten" deformities. Patients may need surgical procedures to cut fingers free. Over time rigid joints and deformities emerge as the damaged skin shrinks and tightens muscles and tendons, causing **contractures**, which further reduces mobility.

In healthy skin, anchoring fibrils made mostly of **Type VII collagen protein** knit the thin epidermis to the dermis below. In RDEB, any of 200 mutations prevent the formation of the fibrils. Gene therapy may offer hope for repairing these mutations.

Gene therapy uses viruses to deliver functional **COL7A1 genes**, which encode the collagen, into cells taken from patients and growing outside the body ("ex vivo"), and then injects the doctored self-cells into selected areas of skin.

RDEB gene therapy is localized, and would coincide with patients' routine hospitalizations, treating one skin area at a time. In younger children with RDEB, wounds start emerging on the body before they become immobile with contractures by their early teens. One of the clinical strategies is to treat earlier and prevent these small areas from becoming larger areas. Focusing on certain areas, like the fingers,may better improve quality of life.

**Fibrocell,** a cell and gene therapy company, is targeting **fibroblasts**, and described the effect of their product on five wounds treated in three adults. At the 12-week mark four of the wounds were greater than or equal to 70% healed and type VII collagen produced, although anchoring fibrils were not yet observed.

1. What part of the body is affected by **epidermolysis bullosa**?

2. How is the disease currently treated?

3. What is a "contracture?"

4. Describe the role of **Type VII collagen** in skin?

5. *Use the graphic on the next page for a more detailed examination of how the process works.* What is the source of the modified fibroblasts?

How is the correct gene added to the cells?

How are the fibroblasts modified?

How are the modified cells then used to treat EB?

6. Consider the underlined sentence in the second to last paragraph. What does this mean for the success of the treatment?

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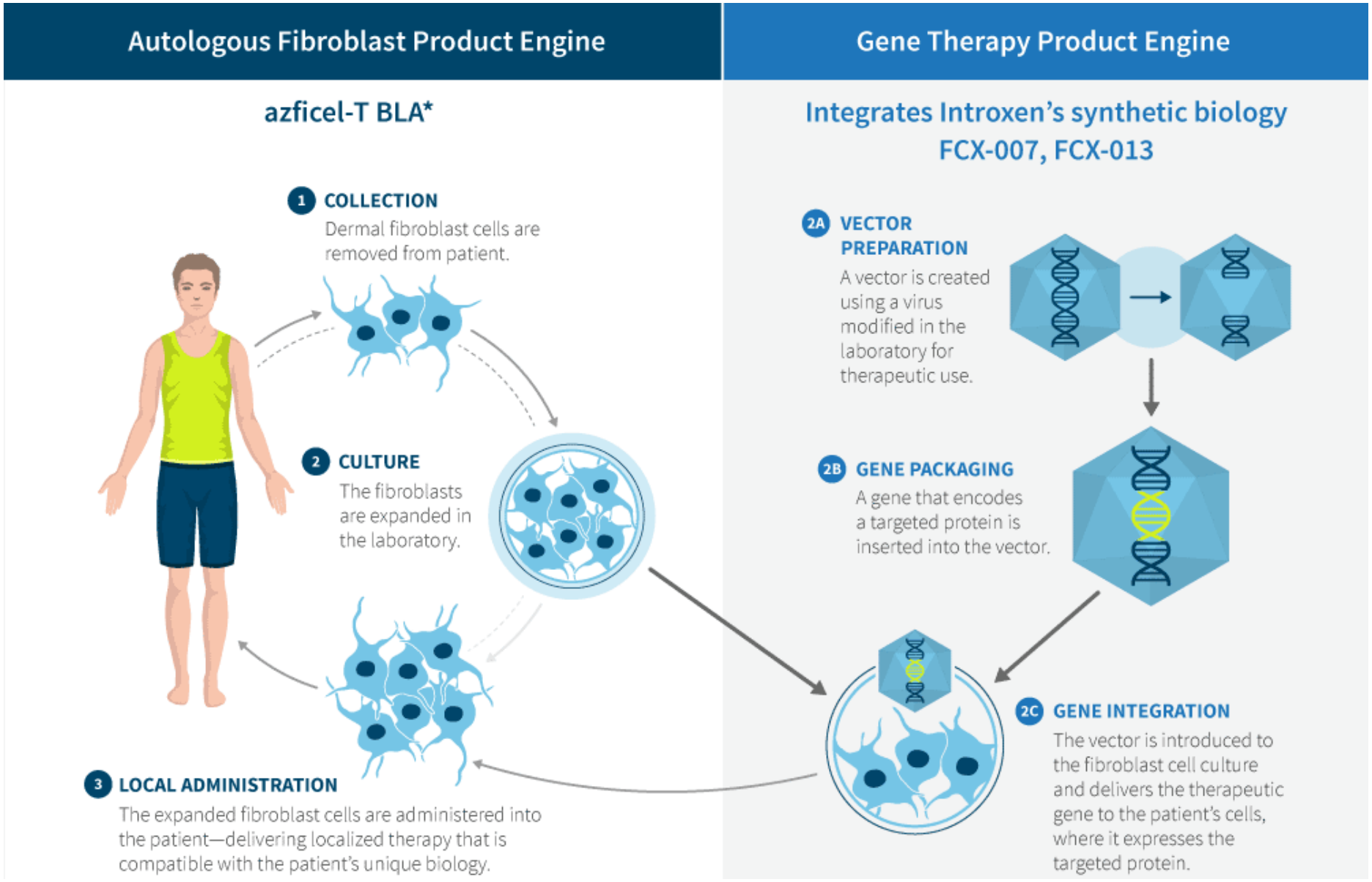


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