Ichthyosis with confetti

Ichthyosis with confetti is a disorder of the skin. Individuals with this condition are born with red, scaly skin all over the body, which can be itchy in some people. In childhood or adolescence, hundreds to thousands of small patches of normal skin appear, usually on the torso. The numerous pale spots surrounded by red skin look like confetti, giving the condition its name. The patches of normal skin increase in number and size over time.

In addition to red, scaly skin, people with ichthyosis with confetti typically have abnormally thick skin on the palms of the hands and soles of the feet (palmoplantar keratoderma). Many affected individuals have excess hair (hirsutism) on some parts of the body, particularly on the arms and legs. Because of their skin abnormalities, people with ichthyosis with confetti are at increased risk of developing skin infections.

Frequency

Ichthyosis with confetti is a rare disorder. Fewer than 20 affected individuals have been described in the medical literature.

Genetic Changes

Mutations in the \textit{KRT10} gene cause ichthyosis with confetti. This gene provides instructions for making a protein called keratin 10, which is found in cells called keratinocytes in the outer layer of the skin (the epidermis). In the fluid-filled space inside these cells (the cytoplasm), this tough, fibrous protein attaches to another keratin protein (produced from a different gene) to form fibers called intermediate filaments. These filaments assemble into strong networks that provide strength and resiliency to the skin.

\textit{KRT10} gene mutations associated with ichthyosis with confetti alter the keratin 10 protein. The altered protein is abnormally transported to the nucleus of cells, where it cannot form networks of intermediate filaments. Loss of these networks disrupts the epidermis, contributing to the red, scaly skin. However, in some abnormal cells, the mutated gene corrects itself through a complex process by which genetic material is exchanged between chromosomes. As a result, normal keratin 10 protein is produced and remains in the cytoplasm. The cell becomes normal and, as it continues to grow and divide, forms small patches of normal skin that give ichthyosis with confetti its name.

Inheritance Pattern

Ichthyosis with confetti is considered to have an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause
the disorder. Usually, the condition is caused by a new mutation that occurs very early in embryonic development (called a de novo mutation). In these cases, the affected individuals have no history of the disorder in their family. In some cases, an affected person inherits the mutation from one affected parent.

Other Names for This Condition
- congenital reticular ichthyosiform erythroderma
- CRIE
- ichthyosis variegata
- IWC

Diagnosis & Management

Genetic Testing
- Genetic Testing Registry: Erythroderma, ichthyosiform, congenital reticular

Other Diagnosis and Management Resources
- Foundation for Ichthyosis and Related Skin Types (FIRST): Skin Care Tips
  http://www.firstskinfoundation.org/skin-care-tips

General Information from MedlinePlus
- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus
- Health Topic: Skin Conditions
  https://medlineplus.gov/skinconditions.html
Educational Resources

- Disease InfoSearch: Erythroderma, ichthyosiform, congenital reticular
  http://www.diseaseinfosearch.org/Erythroderma%2C+ichthyosiform%2C+congenital+reticular/8358
- MalaCards: erythroderma, ichthyosiform, congenital reticular
  http://www.malacards.org/card/erythroderma_ichthyosiform_congenital_reticular
- Merck Manual for Healthcare Professionals: Ichthyosis
  https://www.merckmanuals.com/professional/dermatologic-disorders/cornification-disorders/ichthyosis
- Orphanet: Congenital reticular ichthyosiform erythroderma
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=281190

Patient Support and Advocacy Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST)
  http://www.firstskinfoundation.org/index.cfm
- National Organization for Rare Disorders (NORD): Ichthyosis
  https://rarediseases.org/rare-diseases/ichthyosis/
- Resource List from the University of Kansas Medical Center: Ichthyosis
  http://www.kumc.edu/gec/support/ichthyos.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ichthyosis+with+confetti+5BTIAB%5D%29+OR+%28crie%5BTIAB%5D%29+OR+%28iwc%5BTIAB%5D%29+OR+%28congenital+reticular+ichthyosiform+erythroderma%5BTIAB%5D%29+OR+%28ichthyosis+variegata%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22+AND+373149

OMIM

- ERYTHRODERMA, ICHTHYOSIFORM, CONGENITAL RETICULAR
  http://omim.org/entry/609165

MedGen

- Erythroderma, ichthyosiform, congenital reticular
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9892962

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21929535

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20798280
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3085938/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12636020


Reviewed: February 2014
Published: May 22, 2018

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services