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PICTORIAL ESSAY

# Harlequin Ichthyosis: A Rare Case of Congenital Ichthyosis

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We report a case of Harlequin ichthyosis (HI), extremely rare genetic disorder (1 in 300,000) with high (25%) recurrence rate in successive pregnancies. A preterm baby, born out of a consanguineous marriage, had all the features of HI (Figs. 1, 2). The underlying defect involves mutations in ATP binding cassette (ABCA12) gene responsible for transporting lipids to epidermal cells of skin.

Note thick and fish-like scaly skin on the entire body separated by deep fissures, flat fontanel, ectropion, flattened nose, small nostrils, small ears, eclabium with open mouth, absent eyebrows, eyelashes, scalp hair, swollen extremities resulting in limitation of the joint mobility. There is no definitive treatment for HI other than supportive care. The newborn expired after 7 h in the neonatal ICU.

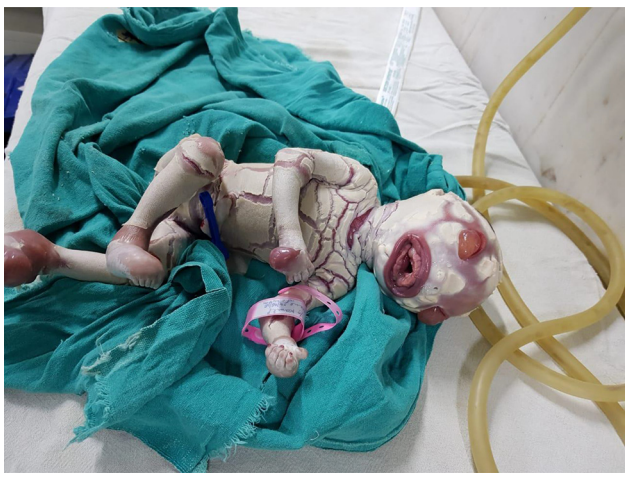

**Fig. 1** Newborn with Harlequin ichthyosis

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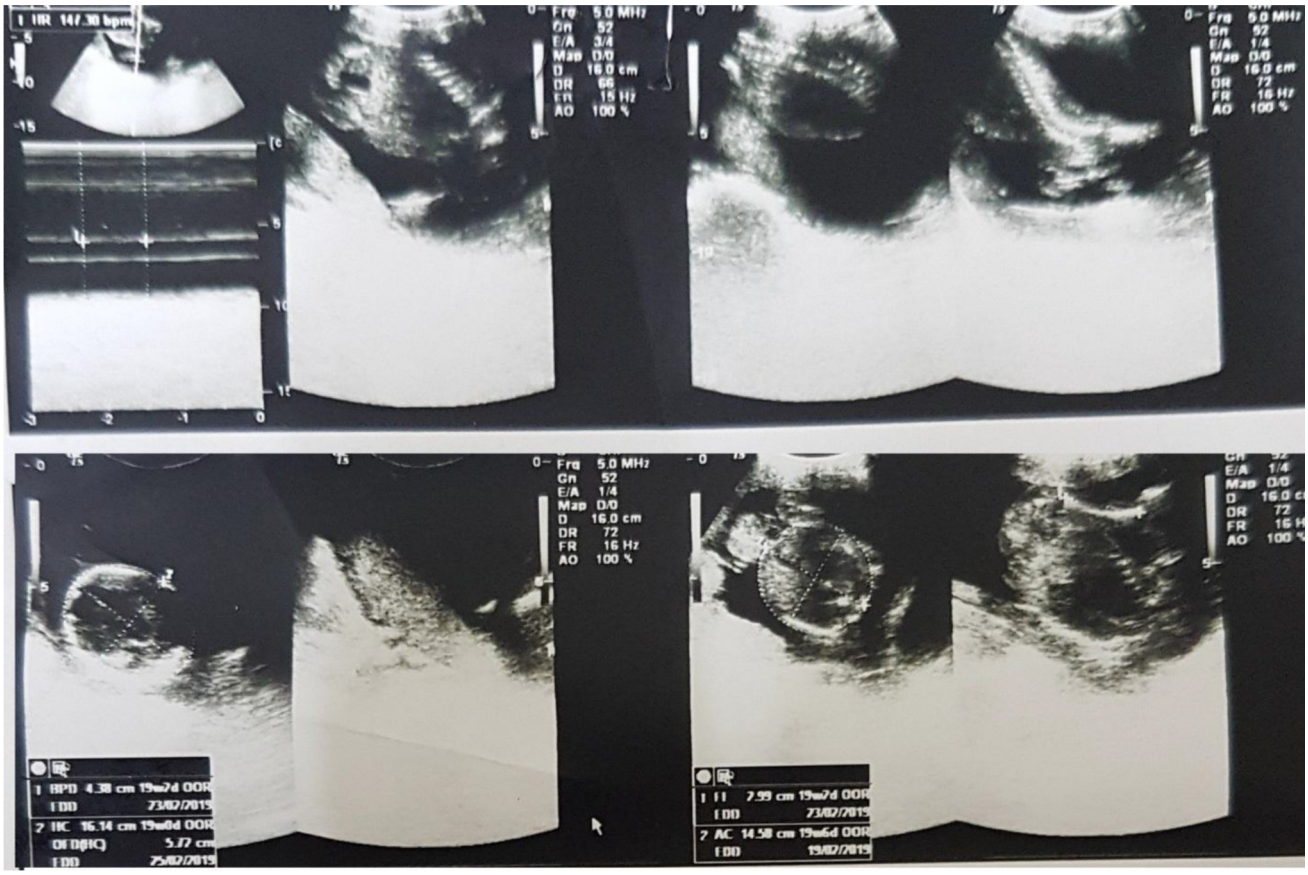

**Fig. 2** Antenatal Ultrasonographic (USG) scan at 17 weeks showed no gross anomaly

## Compliance with Ethical Standards

**Conflict of interest** Dr. Sonia Kataria and Dr. Sangeeta Nangia Ajmani declare that there is no conflict of interest.

**Informed Consent** Informed consent was taken from the patient's parents.

**Research Involving Human Participants and/or Animals** This study does not involve any research conducted on humans or animals.

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## About the Author

**Dr. Sonia Kataria** is working as a Specialist in the department of obstetrics and Gynecology, Kasturba Hospital, Delhi University. She is actively involved in teaching postgraduate students of Obst and Gynae. She did her post-graduation from SMS medical college, Jaipur, Rajasthan and has clinical experience of 15 years in this field. Her fields of interest include Reproductive medicine and high-risk pregnancies.