

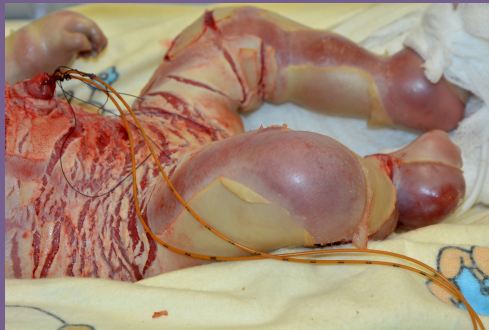
## Harlequin ichthyosis- case report ID 3587

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### Objective

Ichthyoses are keratinization disorder that include a group of heterogeneous entities characterized by extreme dryness of the skin. Harlequin ichthyosis is the most severe form of autosomal recessive congenital ichthyoses, with prevalence of 1:300 000-1:1000 000. The disease is caused by mutation in ABCA 12 gene, which is involved in lipid transport to the skin. The main phenotype is dry scaly fish-like skin, with deep erythematous fissures between thick yellowish armor-like plaques involving entire body surface, eversion of the superior and inferior eyelids (ectropion), both lips (eclabium or fish mouth appearance).



Pict.1,2,3 Postpartum images

### Case report

36-year old multipara was admitted to our Unit in 36 weeks of gestation with spontaneous labor. Due transverse position of the fetus a cesarean section was performed. We delivered a baby with phenotypic features of Harlequin baby: scaly fish-like skin with deep erythematous fissures involving the entire body surface, the eversion of the superior and inferior eyelids (ectropion) and both lips (eclabium or fish mouth appearance), the absence of eyelashes and eyebrows, sparse hair, abnormal flattened ears and broadened flat nose, microcephaly, abnormally fixed limbs and fingers, and toes in rigid flexion. No prenatal diagnosis of this condition was made. Newborn died in a 2 day of life.

### Conclusion

Prenatal diagnosis of Harlequin ichthyosis is extremely difficult in low- risk patients (ie, patients with no family history). Most of the characteristic features will only become apparent later in pregnancy when restricted skin development becomes a limitation to fetal growth and movements. Even at third-trimester US examination, if there is no suspicion, it is quite hard to diagnose this entity. Due to risk of subsequent pregnancy a patients should be offer a preimplantation genetic diagnosis.