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Supplemental Data

Neu-Laxova Syndrome, an Inborn Error of Serine Metabolism, Is Caused by Mutations in *PHGDH*

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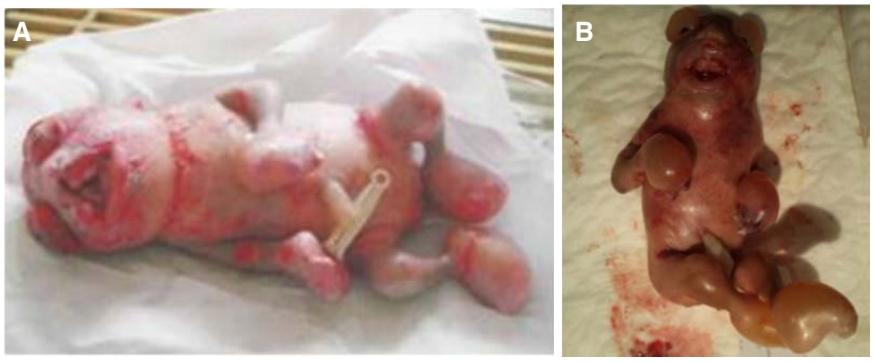

Figure S1. (A) Postnatal photograph of the index from Family 2 showing severe microcephaly, micrognathia, bulging eyes with absent eyelids, severe ichthyosis of the skin, a huge cleft lip and palate on the right side, flat nose, short neck, and generalized edema. Also seen are hypoplastic forearms and no discernible digits in the upper/lower limbs. (B) Postnatal photograph of the index from Family 2 showing massive body swelling with marked disfigurement of the face and limbs that appeared engulfed by a thin and shiny membrane.