# Utilizing Whole Exome Sequencing Reveals a Rare Inherited Variant in ABCA3 Gene

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**Abstract**

Respiratory distress syndrome (RDS) is one of the most common neonatal diseases causing early life morbidity and mortality. We present a case of a full-term baby born to consanguineous parents who died due to severe progressive respiratory failure (PRF). Whole exome sequencing (WES) identified a homozygous disease-causing variant in the ABCA3 gene. Histopathological and electron microscopic examination of postmortem lung tissue revealed characteristic findings consistent with congenital surfactant deficiency along with the ultrastructural evidence of 'fried-egg' like inclusions. Our study provides thorough clinical, radiological, and ultrastructural analysis of the variant's clinical impact and elucidates WES's valuable role in the molecular diagnosis of (PRF).

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