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CASE REPORT

Early-Onset Wolman Disease in an Infant with Novel Compound Heterozygous *LIPA* Variants

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ABSTRACT

Background: **Wolman disease is a rare autosomal recessive lysosomal storage disorder caused by lysosomal acid lipase deficiency, with fewer than 150 cases reported. It often presents in infancy with hepatosplenomegaly and adrenal calcification.**
Methods: **We report a 2-month-old female with abdominal distension, vomiting, hepatosplenomegaly, and bilateral adrenal calcification.**
Results: **Laboratory findings showed anemia, thrombocytopenia, hypoalbuminemia, and elevated liver enzymes. Genetic testing revealed novel compound heterozygous *LIPA* variants: a paternal frameshift (c.731_732del, p.Gly244Aspfs*24) and a maternal missense (c.605C>G, p.Pro202Arg).**
Conclusions: **This case expands the *LIPA* variant spectrum and emphasizes the importance of combining clinical data for early diagnosis and counseling.**
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Supplementary Data

Table S1. Hyperlipidemia-related genes tested in this patient.

ABCA1	***ABCG5***	***ABCG8***	***AGPAT2***	***ALMS1***	***ANGPTL3***
APOA1	***APOA5***	***APOB***	***APOC2***	***APOE***	***APPL1***
APTX	***BSCL2***	***CAV1***	***CAVIN1***	***CEL***	***CETP***
CIDEC	***CYP27A1***	***DYRK1B***	***GALNT2***	***GCK***	***GPD1***
GPIHBP1	***HNF1A***	***HNF1B***	***HNF4A***	***INS***	***KLF11***
LCAT	***LDLR***	***LDLRAP1***	***LIPA***	***LIPC***	***LIPE***
LMF1	***LMNA***	***LMNB2***	***LPL***	***MTTP***	***NEUROD1***
PCSK9	***PDX1***	***PLIN1***	***PNPLA2***	***POLD1***	***PPARG***
SAR1B	***ZMPSTE24***				