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【 PICTURES IN CLINICAL MEDICINE 】

LIG4 Syndrome Associated with Hypocellular Myeloid Dysplasia

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Key words: LIG4 syndrome, hypocellular myeloid dysplasia, double-strand break repair

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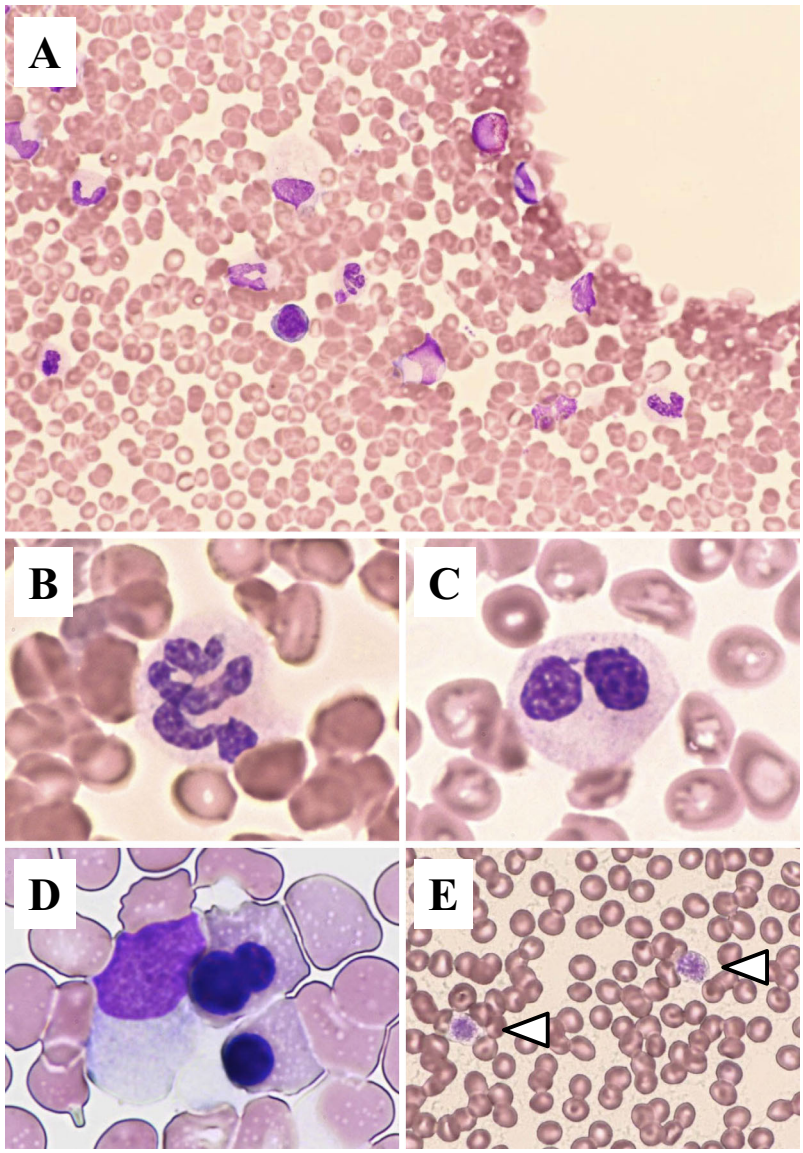

Picture.

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A 17-year-old boy with microcephaly, short stature, pancytopenia and hypogammaglobulinemia was referred to our department. An exome analysis was performed because of suspected congenital immunodeficiency. Based on the results, he was definitively diagnosed with LIG4 syndrome, an extremely rare condition (1). His bone marrow smear performed at the initial examination demonstrated hypocellularity (nucleated cell count of 8,000/μL; Picture A), myeloid lineage dysplasia (Picture B and C), erythroid lineages dysplasia (Picture D), and giant platelets (Picture E, arrowheads). Furthermore, there were no morphological changes in the lymphoid lineage cells. LIG4, an enzyme that plays a crucial role in double-strand break (DSB) repair, is closely involved in lymphocyte differentiation and hematopoietic stem cell maintenance. Various single-nucleotide polymorphisms related to DSB repair, including LIG4, have been reported in cases with myelodysplastic syndrome (MDS) according to a genome-wide analysis, which suggests a close relationship between MDS pathogenesis and mutations in DSB repair (2). To our knowledge, this is the first report of a patient with LIG4 syndrome exhibiting hypocellular myeloid dysplasia.

The authors state that they have no Conflict of Interest (COI).

References

1. Tamura S, Higuchi K, Tamaki M, et al. Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. Clin Immunol **160**: 255-260, 2015.
2. Ribeiro HL Junior, Soares Maia AR, Costa MB, et al. Influence of functional polymorphisms in DNA repair genes of myelodysplastic syndrome. Leuk Res **48**: 62-72, 2016.