Primary immunodeficiency caused by mutations in RIPK1: report of three case

R.Belbouab , S.Hakem, M.Bounaas, Y.Ferhani, S.Sokhal,R.Boukari
Department of Pediatrics, Mustapha University Hospital, Algiers.

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Background

Mutations in RIPK1 (receptor-interacting serine/threonine kinase 1) were associated with impaired NF-kB activity, defective differentiation of T and B cells, increased inflammasome activity, and reduced response to TNFR1-mediated cell death in intestinal epithelial cells.

Purpose:

Describe the clinical expression of three children of the same siblings presenting with mutation in RIPK1

Methods:

The data was collected from the medical records of patients during their hospitalization and their follow-up in our department.

Results:

We report the case of three children, two boys and a girl, all members of the same family whose parents are consanguineous.
the onset of the disease started very early in life (6, 1 and 3 months) the clinical symptoms were similar for the three patients marked by gastrointestinal manifestations suggestive of VEO-IBD made up of watery diarrhea, abdominal pain, oral aphthous lesions, anal fistulas, growth retardation.

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ulcerative colitis on endoscopy (A1L2B1 according to the Paris classification) and chronic inflammation on histology. For extra intestinal manifestations, the three patients presented several infections, in particular pneumonia and purulent otitis, two patients presented attacks of tetanus.

The diagnosis was made late at the age of 10, 6 and 3 years by molecular biology (D.Kotlarz, University Hospital LMU Munich)

Therapeutic management consisted of antibiotic combined with antifungal therapy for the three patients as well as azathioprine treatment in one patient.

Progress under treatment led to a reduction in diarrhea and infections.

Conclusion:

It is important to identify patients with monogenic Inflammatory Bowel Diseases (IBD) as management may differ from classical IBD. Genetic screening for monogenic IBD is recommended in all patients with very early-onset IBD (<6 years), in particular, in those patients with relevant comorbidity, extraintestinal manifestations, and/or family history. RIPK1 Deficiency Causes Immunodeficiency and Intestinal Inflammation.

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WHERE AND HOW TO FIND THEM?
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