Interleukin 10 receptor gene mutations in very early–onset inflammatory bowel disease: presentation of two cases

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Objective
Role of genetic aetiology in the management of rare clinical condition.

Methods
In the aetiology of very early onset inflammatory bowel disease (IBD), genetic defects play a greater role than environmental factors. Mutations in interleukin-10 receptor (IL-10R) genes are one of the cause of very early-onset inflammatory bowel disease with perianal lesions. There are only few cases reported in literature so far. We identified mutations affecting the IL-10 pathway in 2 patients with a diagnosis of very early onset IBD. The presentation resembled crohns disease in terms of clinical course, and anatomic and histopathological involvement; however, response to conventional treatment was particularly poor. Intractable enterocolitis, failure to thrive, perianal disease, and fistula formation was noted. Haemopoietic stem cell transplant was carried out in both the patients with IL-10 deficiency. This proved to be an effective therapy in both cases leading to rapid improvement in clinical symptoms and quality of life. However one case succumbed to sepsis. Genetic counselling of the family was conducted and prenatal diagnosis was offered for future pregnancies with successful outcome.

Results
In conclusion, our observation shows that IL-10/IL-10R deficiencies are primary immunodeficiencies that need to be considered in patients with therapy-refractory early-onset IBD. Both children were born to consanguineous parents and had a positive family history.

Conclusion
Clinical outcome can be improved if genetic aetiology of very early onset IBD is identified. Genetic counselling can be offered regarding prenatal diagnosis in future pregnancies.