A rare case of laryngeal granulomatous inflammation caused by RAG1-Deficient Severe Combined Immunodeficiency (SCID)
Katrina Mason, Claudia Nogueira, David Royan, Mark Felton, Victoria Possamai
Evelina Childrens Hospital, London, UK

- Differentials for laryngeal granulomatous conditions in the paediatric populations include:
  - Granulomatosis with Polyangitis (GPA)
  - Sarcoidosis
  - Tuberculosis
  - Rare infective processes
- We present the rare case of a 12 year old with extensive progressive laryngeal granulomatosis caused by Rag-1 Deficient Severe Combined Immunodeficiency (SCID)

A 12 year old was transferred to the Evelina Children’s Hospital with a 6-year history of chronic cough and respiratory colonisation with pseudomonas requiring multiple courses of antibiotics. Empirical TB treatment was started despite negative cultures for AFB.

The persistent cough and hoarseness was investigated with Microlaryngoscopy & bronchoscopy (MLB). This showed diffuse laryngeal granulation which histologically showed granulomatous inflammation with dense chronic inflammatory infiltrate (lymphocytes, histiocytes, epitheliod granulomas). Over a few weeks despite bing on steroids her laryngeal function deteriorated and she developed dysphagia and stridor. MLB with tracheostomy was required which showed clear progression with dense granulation affecting the entirety of the larynx.

A whole body MRI revealed widespread bone and lung lesions with bronchiectasis. Haematological analysis showed an abnormal immunological profile with low B cell/CD4, normal NK cell and monoclonal T cell expansion. This was eventually confirmed as RAG1 Severe combined Immunodeficiency (SCID).

The patient has been transferred to GOSH for bone marrow transplant

What is SCID?
Severe Combined Immunodeficiency, is a syndrome caused by mutations in different genes whose products are crucial for development and function of both T and B cells. The clinical presentation usually includes severe recurrent and potentially lethal infections in early infancy. Disease can be controlled through long-term antibiotics and antifungals and through treatment of opportunistic infections, but it can prove fatal. Cure is offered through bone marrow transplant if possible.