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Progressive gait instability in a young girl; what will happen at the end?

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DESCRIPTION

An 8-year-old girl had been experiencing a progressive impairment in stance and gait since the age of 20 months and a diagnosis of ataxic cerebral palsy was given accordingly. She had recurrent upper respiratory tract infections over the past few years. Her mother noted that the patient's eyes were red and her paediatrician told her that this is a chronic allergy (figure 1). The girl demonstrated a good sense of humour and no mental subnormality. All modalities of sensation were intact and both planter reflexes were flexors. The girl sways back and forth and side by side when she stands and walks. The red eyes were actually conjunctival telangiectasias and note the vitiligo as well; the girl has ataxia telangiectasia.

The appearance of these telangiectasia (especially on the bulbar conjunctiva) often allows the diagnosis to be made in a child with ataxia.¹ These telangiectasias usual appear around the ages of 3–6 years (therefore, later than ataxia), are venous in origin and rarely bleed. Because of their persistence and reddish hue, they are commonly mistaken for chronic conjunctivitis or perennial allergy. The most fearful complication is the high risk of developing of a wide range of malignancies, which is 100-fold higher than the general population; Non-Hodgkin's (including Burkitt's) lymphoma accounts for 40% of all malignancies.² The low levels of serum IgA, IgE and IgG subclass 2; decreased levels of CD3 and CD4 T cells; impaired delayed hypersensitivity reactions and thymic hypoplasia are the usual immune system abnormalities of the disease.³

Figure 1 The patient's face.

Learning points

- ▶ The bulbar conjunctiva telangiectasias appear later than ataxia, and therefore, the eyes of all patients with relentlessly progressive instability of stance and gait should be carefully scrutinised.
- ▶ The impaired immune system and surveillance in ataxia telangiectasia predispose patients to recurrent upper and lower respiratory tract infections and a multitude of malignancies; the most fearful complications of the disease.
- ▶ In contrast to what is expected, children with ataxia telangiectasia are socially responsive, appreciative and non-demanding, and that mental subnormality is not a feature of the disease.

Competing interests None.

Patient consent Obtained.

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