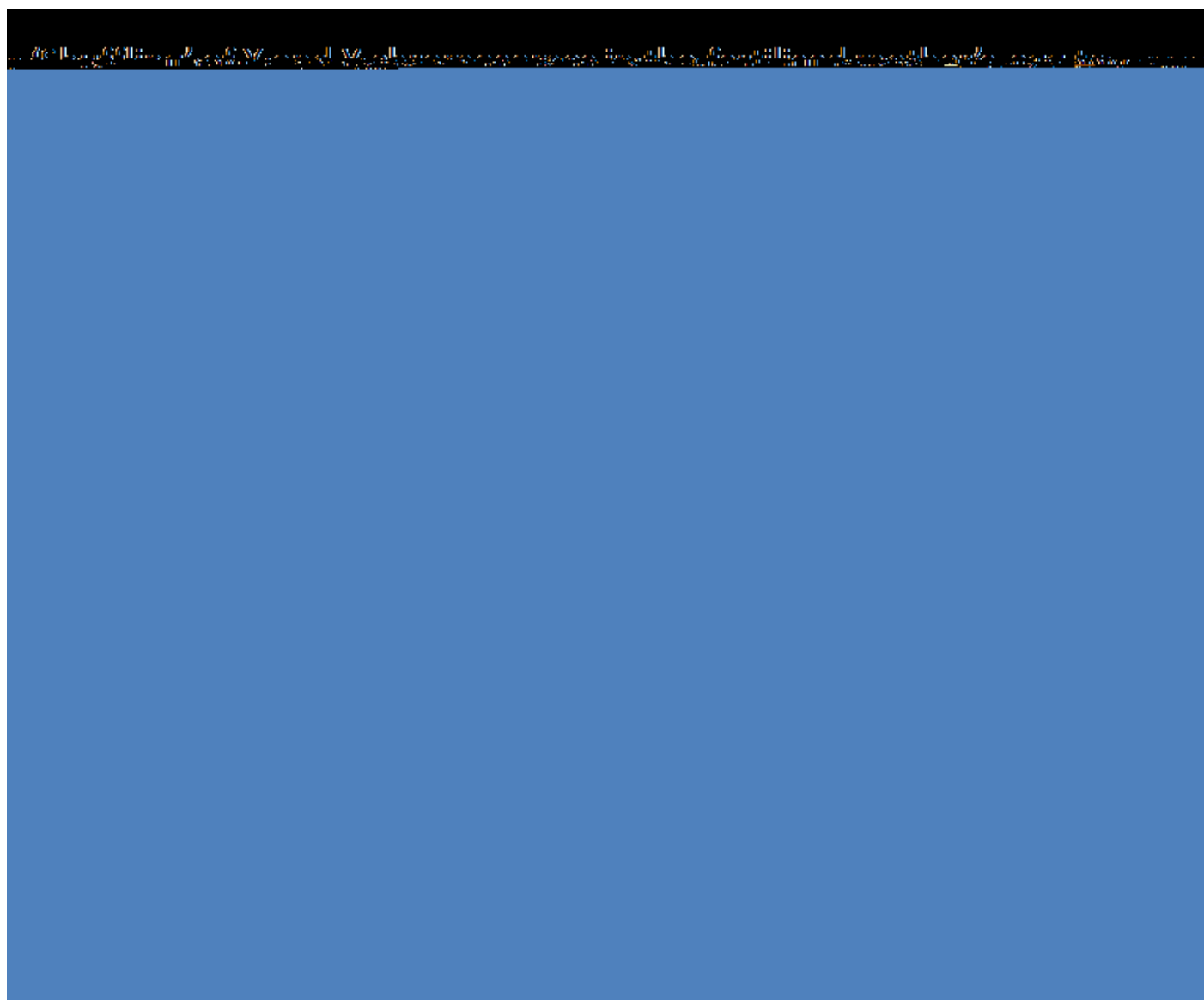


X-linked agammaglobulinaemia XLA

Background by Dr David Webster

XLA patients are susceptible to a well-recognised list of infections that cause problems in the sinuses and lungs, the bowel and less frequently the joints, bladder or brain. Some of these infections are easy to diagnose while others require tests done in special laboratories before the appropriate treatment can start. Fortunately most of the difficult to diagnose infections have become much rarer in XLA patients living in western countries over the past 20 years because the standard dose of immunoglobulin therapy has been increased. Nevertheless there is no room for complacency and patients should have confidence that their specialist has the necessary experience of XLA to know what tests are needed. In general, the earlier in life immunoglobulin treatment is started the better, as any permanent damage to the lungs from infection will increase the chances of further infection, and damage to joints or brain might be irreversible.

What of the future? XLA should be an ideal disease to cure with gene therapy or bone marrow stem cell transplantation. Although major advances are being made in correcting gene defects or replacing bone marrow in more severe and life-threatening PIDs, these procedures still carry too much risk for most XLA patients who generally do well if managed properly. However we may take a very different view in 10 years time.



X-linked agammaglobulinaemia (XLA)

I was diagnosed with XLA in 1993 when I was in my early forties. My job as a manager at my local hospital brought me into daily contact with medical staff and one day a Chest Physician pointed out that I seemed to suffer from a cough and cold almost continuously which was not normal, so he arranged for me to see other consultants for tests. You should remember that XLA was not recognised until the 1950s and is so rare that only a few GPs will ever encounter it. I was treated with antibiotics and told that I had a weak chest and should take care of myself! However, I became increasingly aware that the infections were getting worse and I was taking longer to recover. I almost missed my wedding as I went down with flu just days before.

When I was given the results of the tests, I did not understand what I was being told. I only heard the words Immune Deficiency and immediately thought I had contracted AIDS. Many of you will remember that in the late 80s and early 90s AIDS was constantly in the headlines as the new