

HEREDITARY ANGIO-OEDEMA

This is a rare genetic condition also called hereditary angio-oedema. In this condition there is a deficiency in the blood of a factor which controls the level of inflammation in the body - C1 inhibitor. Lack of this factor results in spontaneous activation of the complement system resulting in episodes of localised swelling of the tissues, which can be very painful. Swelling may also occur in the lining of the gut resulting in abdominal pain often accompanied by vomiting which may be misdiagnosed as appendicitis. If the swelling occurs in the wrong part of the body, for instance the throat, the attack can be life threatening. In patients suffering frequent attacks prophylaxis to try and reduce the frequency of these attacks is used. In children this usually comprises Tranexamic acid and in adults retarded anabolic steroids such as Danazol or Stanazol. While many attacks are without obvious provocation, we do know that there are certain things that can increase the likelihood of an attack and these include trauma (including minor surgery and dental treatment), emotional upset and stress.

Emergency treatment of an attack is necessary if the episode is affecting the face, mouth, neck region and therefore threatening the upper airway. This comprises an infusion of purified C1 inhibitor concentrate (C1 inhibitor TIM3 preparation). The family should be provided with a vial of this and told to take it along when seeking urgent medical attention. The dose is age dependant. For those under 10 years of age 500 units is given and for those over 10 years of age 1000 units. The inhibitor will normally terminate the attack very rapidly. It is also used for attacks causing severe acute abdominal pain and vomiting if these attacks are not controllable with simple measures. The concentrate should also be used prophylactically to cover minor surgical procedures including dental treatment and tonsillectomy.

Further information on this condition can be obtained from the Primary Immunodeficiency Association and, in an emergency, advice can be sought by telephoning the number given below during office hours or contacting the Duty Immunology Registrar (via the Great Ormond Street Hospital switchboard) out of hours.

Yours sincerely,

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