

BSUH Paediatric Guidelines

Protocol for immunological investigation of 22q deletion

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INTRODUCTION

The immune defect associated with 22q can be anything from mild and insignificant, to so severe they act like a baby with SCID and need a thymic transplant. This protocol has been discussed with Dr. Burns, Dr Davies and Dr. Alison Jones, Consultant Immunologists at Great Ormond Street.

AT DIAGNOSIS

1. Full lymphocyte subsets to look at T cells and B cells
2. Memory T cell panel (to look at naïve T cells to give an indication of thymic function)
3. Immunoglobulins,
4. Vaccine responses (if old enough to have vaccines), particularly pneumococcal with vaccine challenge if necessary
5. T cell function with a PHA stimulation test.

FOLLOW UP

Lymphocyte subsets and immunoglobulins if abnormal: yearly or every couple of years depending on infection history.

If normal: just repeat once after five years.

PHA: If PHA is normal at baseline it does not need repeating, if abnormal will need intermittent FU depending on severity.

TREATMENT

If immunoglobulins are very low: patient may rarely need replacement IG

If no specific vaccine responses: boost with vaccines. May need Azithromycin prophylaxis or rarely immunoglobulin replacement.

T cell dysfunction: harder to treat- depends on viral illnesses and T cell numbers.

Please discuss abnormal results with paediatric Immunology (KF at RACH and / or immunology team at GOSH)

Live Vaccine: if low T cells please discuss but generally we use HIV guidelines.

Doing Specific Tests!!

We can do all the bloods here at RACH apart from the PHA and memory T cell panel which we would send to Great Ormond Street.

PHA and Memory lymphocytes panel

These can be done at Great Ormond Street Hospital, Tuesday, Thursday or Friday. The blood should be taken early morning in Brighton and couriered up to get there by 2 pm at the latest. Please ring first; 0207 405 9200 Ext. 8835 to arrange day and test.

PHA need 1 ml lithium heparin from patient and 1 ml lithium heparin from a healthy control (any young adult happy to be bled).

Memory lymphocytes panel need 1 ml EDTA

Blood should be sent for the attention of: Dr. Kimberley Gilmore, Immunology Lab, Great Ormond Street Hospital, Great Ormond Street, London WC1N 3JH.