CARPENTER SYNDROME (ACPS II) – HSS Service
OMIM 201000

INTRODUCTION
Carpenter syndrome (also known as acrocephalopolysyndactyly type II (ACPS II)) is a rare autosomal recessive disorder.

The characteristic manifestations of ACPS II are craniosynostosis (most commonly of metopic or sagittal sutures), obesity, polydactyly and soft tissue syndactyly. Other well-recognised features include brachydactyly with shortening or absence of the middle phalanges, molar agenesis, genu valgum, hypogenitalism, congenital heart defects, umbilical hernia and learning difficulties.

The main benefit of molecular testing is confirmation of the diagnosis, which then enables carrier testing in parents and family members, and prenatal diagnosis may also be available to families.

The types of mutations that have been reported in ACPS II have been mainly nonsense, missense, and frameshift mutations in RAB23.

TESTING AND REFERRALS
All diagnostic samples should be accompanied by a completed clinical questionnaire & consent form (click here)
The aim is to provide a joint Molecular and Clinical service, and Professor Andrew Wilkie (+44 (0)1865 222619) is available for advice. We test:
  o Clinically affected patients
  o Relatives of clinically affected patients in whom mutations have been identified
  o Prenatal testing is available in cases where two mutations have been identified. These must be discussed with the laboratory and arranged in advance.

REFERRALS
From Clinical Genetics, Paediatrics, Craniofacial clinics, Dysmorphologists. Prenatal testing is only accepted from Clinical Genetics and/or Prenatal Diagnosis.

STRATEGY
Family tests: When mutations have been identified in a family the individual is tested only for the familial mutations.
For new cases: At least 95% are due to mutations of RAB23.

TECHNICAL INFORMATION
Mutation analysis: Bidirectional fluorescent sequencing of the coding regions of RAB23 (exons 2-7).

TARGET REPORTING TIMES: supraregionally funded by HSS for referrals from the UK
Diagnostic: 8 weeks
Familial Mutation test: 10 days
Prenatal testing (includes maternal contamination check): 3 days

N.B. Details are correct for the date of printing only – last updated 11/06/2015