

“Wolfram syndrome syndrome diagnostic & multidisciplinary clinical service” UPDATE Sat 28th Jan 2012

*National Commissioning Group
For Highly Specialised Services*

Birmingham Children's Hospital 
NHS Foundation Trust

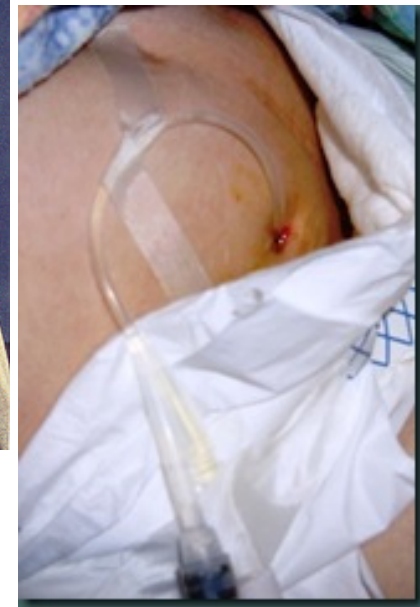
University Hospital Birmingham NHS Foundation Trust

Summary of presentation

- Introduction: who I am, who's who
- Patient and family support organisations
- Clinic support: Spanish, UK service clinics
- The NHS UK specialist clinics

Wolfram Syndrome

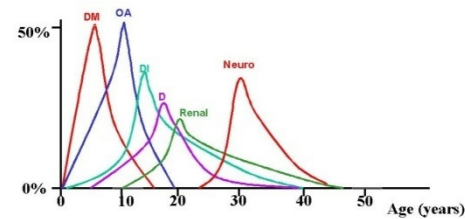
- Optic atrophy
- Insulin dep Diabetes
- Sensorineural deafness
- Diabetes insipidus
- Neuropathic bladder
- Neurodegeneration



Birmingham UK and Wolfram research



Natural history

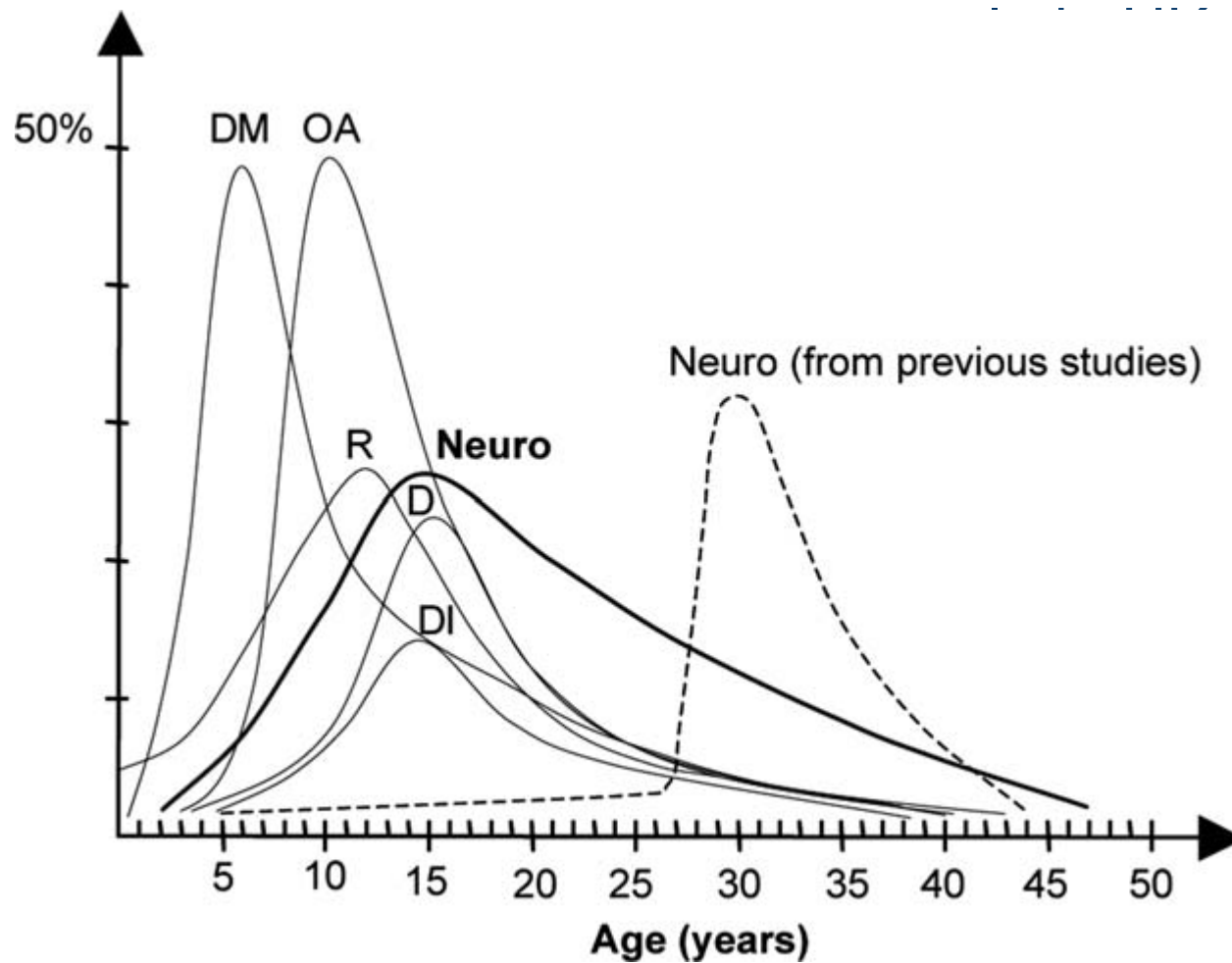


2001
NHS genetic testing
service

*(Barrett et al lancet
1995)*

How common?

1:700,000 children; ~120



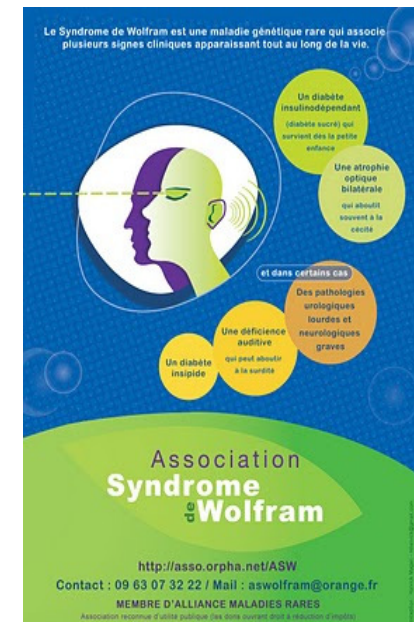
Chausseot A et al Ann Neurol 2010

Patient support (1): groups

- French Wolfram Association
 - Organised international meetings Oct 2009, June 2010, Oct 2011
 - Family meetings
 - Supported by AFM, Telethon
- UK Wolfram support group
 - Support from Wellchild
 - Family meeting Jan 2012



Mdm Nolwenn
Jaffre



the UK's only WS family support web site

Patient support 2: Orphanet: the European portal for info about rare diseases and services

Orphanet - Windows Internet Explorer

http://www.orpha.net/consor/cgi-bin/index.php

File Edit View Favorites Tools Help

Orphanet

Languages : Français | **English** | Español | Deutsch | Italiano | Português

Homepage
Help
Contact us

orphanet
Inserm

Rare diseases Orphan drugs Expert centres Diagnostic tests Research and trials Patient organisations Professionals and institutions Other information

SIMPLE SEARCH

Search a disease **OK**

> Alphabetical list of rare diseases

OTHER SEARCH OPTION(S)

- > Orphan drugs
- > Research and trials
- > Diagnostic tests
- > Patient organisations
- > Expert centres
- > Professionals and institutions

ORPHANET DATA

Diseases	: 5954
Expert centres	: 4942
Laboratories	: 5424
Professionals	: 15019
Daily visitors	: 12810

RARE DISEASES

- > Information about a disease
- > Alphabetical list
- > Search by clinical sign
- > Search by gene
- > Emergency guidelines
- > Encyclopaedia for patients
- > Encyclopaedia for professionals
- > Classifications
- > About Rare Diseases
- > Prevalence of Rare Diseases

RESOURCES DIRECTORY

- > Expert centres
- > Diagnostic tests
- > Research projects
- > Registries / databases

Services for professionals

- > Encyclopaedia for professionals
- > Search by clinical sign
- > Emergency guidelines
- > Register your activity

Services for patients

- > Information about a disease
- > Encyclopaedia for patients
- > Patient organisations
- > Expert centres

NEWS

- > Plan national maladies rares français 2011-2014 (pdf) [Z]
- > Clinical Utility Gene Cards now

There is no disease so **rare** that it does not deserve attention

Rare diseases are rare, but rare disease patients are **numerous**

[About Orphanet](#) | [Quality charter](#)
[Register your activity](#)

Languages : Français | **English** | Español | Deutsch | Italiano | Português

Done, but with errors on page.

start Orphanet - Windows ... Session - Windows In... desktop - Notepad Desktop Manager

Internet 100% 12:55

Clinical support: Spanish service clinic

- Inmaculada-Huerca-Overa hospital in Almeria.
- Coordinated by Dr Gema Esteban-Bueno
- Multidisciplinary team: Endocrinology, Internal Medicine, Psychiatry, Urology, ORL, and primary attention physicians.
- The aim of this unit is to provide the best attention to the Wolfram patients in a complete and coordinated way.
- First clinic for Spanish patients held in April



Visit to St Louis Children's Hospital MDT Wolfram clinic Aug 12/13th 2011

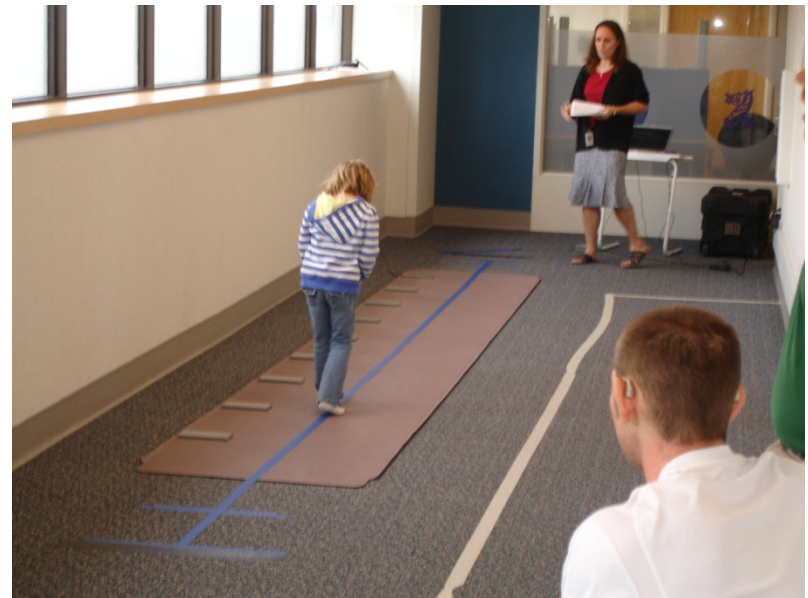


St Louis Wolfram clinic



- 14 children and young people
- Seen over 2 days
- 'research clinic'
- Funded by donations
- Travel self funded

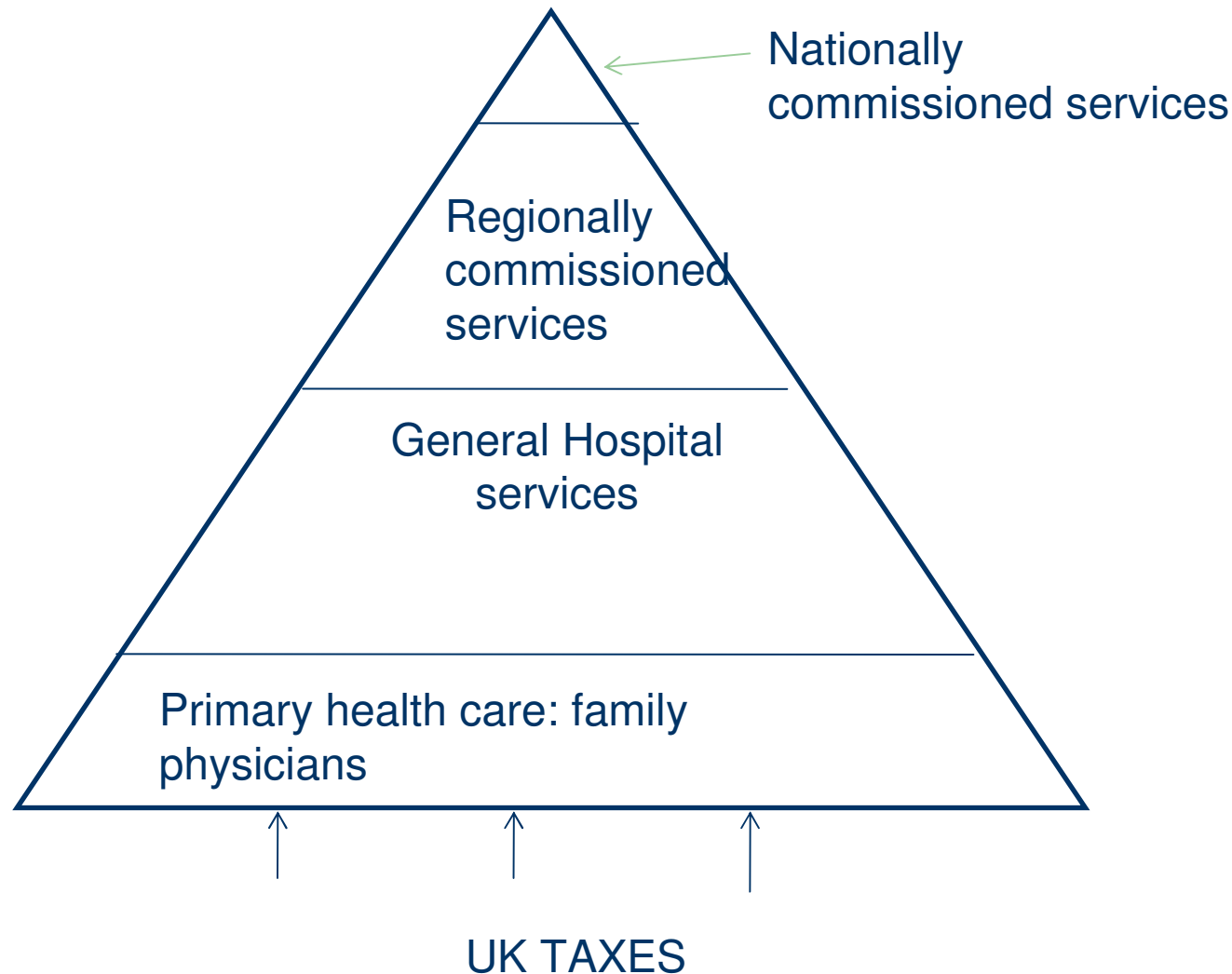
Venue: Clinical research facility



Main lessons learnt

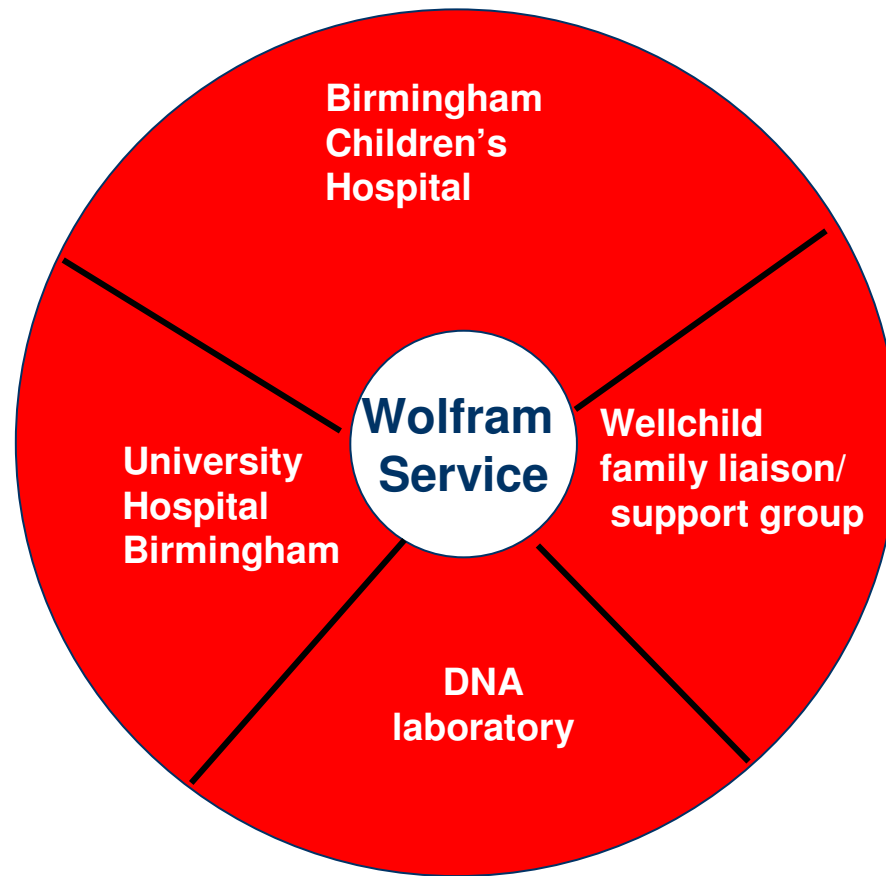
- Get families together before clinic starts
- How to organise fasting bloods on all children at the same time
- Ophthalmology appts difficult to schedule everywhere!
- Importance of timely, explanatory follow-up letter to families
- Vital importance of engaging local physicians: communication
- Need for detailed MRI to chart progress
- Identification of balance problems from a young age

How rare disease services get funded in UK



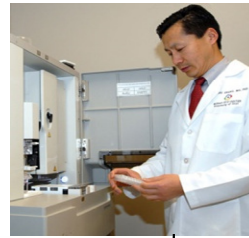
Clinical support: UK Wolfram clinics

- Funded by Government NHS
- National centre on 2 sites
- Key objectives:
 - Monitor and manage disease progression
 - Coordinate care and management of children and adults with Wolfram syndrome
 - Provide expert support for local healthcare providers
 - Establish a disease specific centre of excellence and expertise.
 - To provide national genetic testing service and prenatal diagnosis
- 4 child + 2 adult + 3 outreach clinics/year.



- Outcomes
 - QoI to be maximised as long as possible
 - Hba1c
 - Renal function maximised as long as possible
- Key performance indicators
 - All patients screened for diabetes insipidus two yearly
 - To develop guidelines for patient management
 - All patients over 15 years to be screened for neurological probs
 - Age of diagnosis of referred patients to fall
 - Development of a family outcome tool

The Wolfram MDT clinic



Clinical scientist – molecular genetic testing



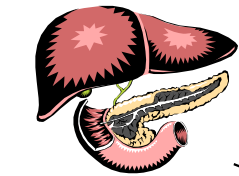
Wellchild Family liaison officer



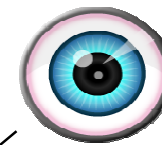
Nurse specialist And Research Nurse



MR Spectroscopy



Endocrine

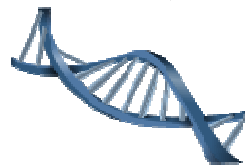


Ophthalmology

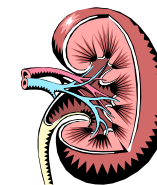


Neurology

Clinical psychology



Genetics and counselling



Urology and urodynamics

UK Wolfram clinic timetable

- Arrive day before
 - Stay Thistle hotel, meet and greet, research consent
- Morning:
 - Fasting bloods if required. Urodynamics, MRI brain, urology assessment
- Lunch:
 - Meet families
- Afternoon
 - Neurology, endocrine, genetics, psychology, audiology, ophth
- Clinic dates Oct 5th, Feb 22nd 2012
- Clinic venue
 - Wellcome Trust Clinical Research Facility
- Individualised appts:
 - Families let us know 6 weeks before if need to see other specialists
 - Dermatology, ENT, Repiratory

The teams

- BCH

- Porter (admin)
- Barrett, Gleeson
- Gupta (Neuro)
- McCarthy (Urology)
- Williams (Genetics)
- Ainsworth (Ophth)
- Clin psychology
- Audiology
- Urodynamics
- (ENT, Resp)

- Wellchild

- Rachel Bates

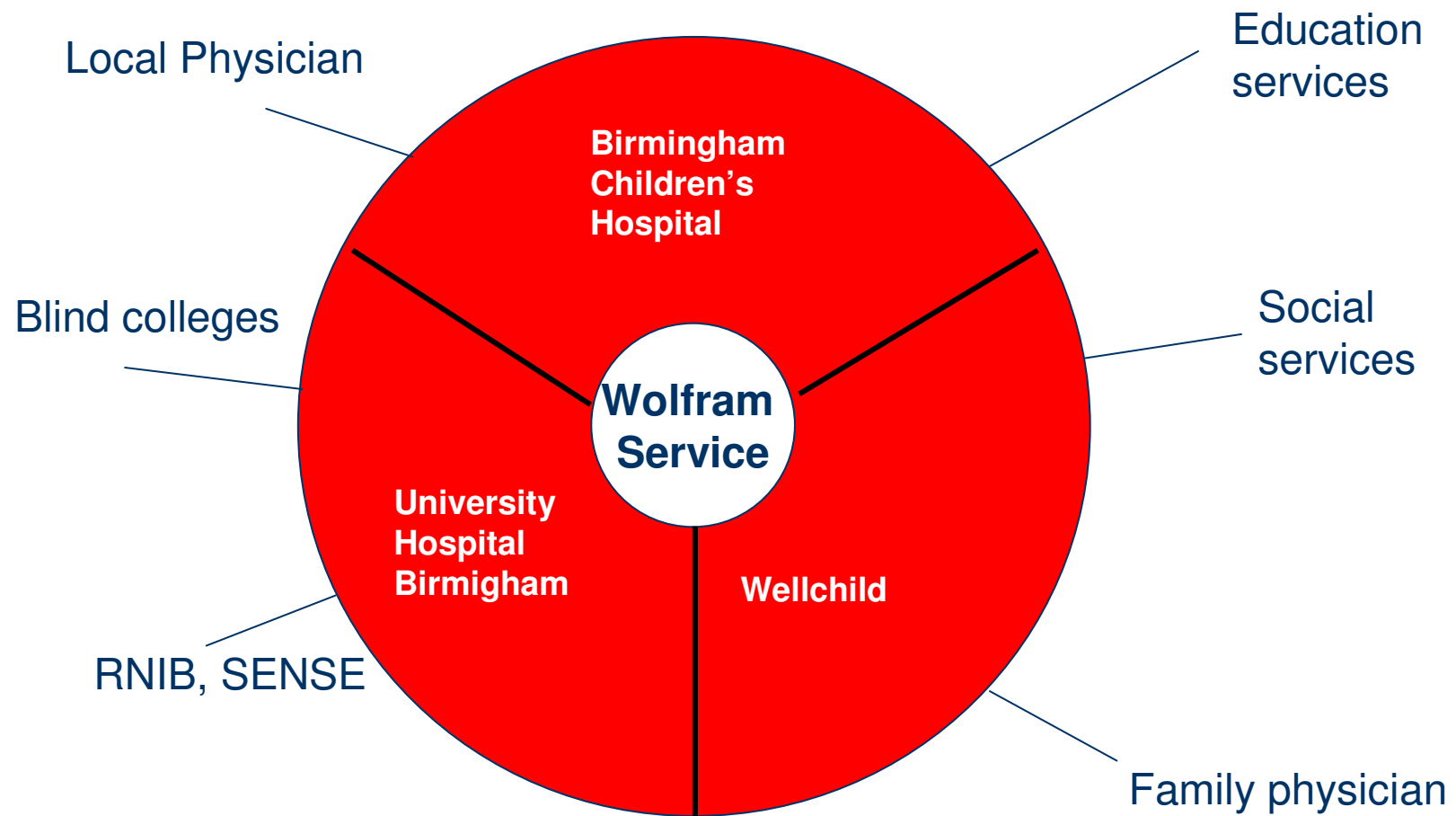
- QEH

- Morrison (neuro)
- Williams (genetics)
- Clin Psychology
- Toogood
(endocrinology)

Timetable

- Patients travel to centre previous evening
- 8.30 Clinical team meets
- 9.00 Clinic starts
 - MRI, urodynamics, audiology, urology
- 12.30 Lunch
- 1.15 Education session
 - living skills, benefits
- 2.15 Clinic recommences
 - Endocrine, neurology, ophthalmology, genetics, psychology
- 4.30 Debrief team meeting

Linking with Community Services for continuity of care



SUMMARY

- Clinics funded for 5 years.
- You need to tell us what you want
- We need family involvement
- Structured service should help when new treatments become available