

BPSU impact - case studies



[British Paediatric Surveillance Unit team](#)

The BPSU has facilitated research into over 100 rare conditions. In many instances, the findings have had an immense influence over decisions relating to the condition and patient. Read case studies on the impact of BPSU's research below.

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Health policy

From its early beginnings, BPSU surveillance data has influenced proposals in many areas of health policy. Following the establishment of the National Screening Committee in the early 1990s, the BPSU was seen as the perfect conduit for gathering evidence to help decide whether national screening on a number of conditions, particularly metabolic conditions, should start, end or continue.

Decisions on health policy are also made by the Joint Committee on Vaccinations and Immunisation (JCVI) and by the Medicines Health Regulatory Authority (MHRA) using BPSU evidence.

Metabolic

Medium chain acyl co-A dehydrogenase deficiency (MCADD) - MCADD is a rare genetic condition in which an individual has problems breaking down fatty acids for energy. It affects around 1 in 10,000 babies born in the UK each year, and is life threatening if not discovered

early. Two BPSU facilitated studies were undertaken by [UCL Great Ormond Street Institute of Child Health](#) in 1994 and 2004. Following the 1994 study, a recommendation was made to the NSC to pilot a screening. In February 2007, the Secretary of State for Health announced that screening of MCADD was to be added to the newborn bloodspot screening programme in England in a phased roll out. The BPSU were able to assist in monitoring the reliability of the screening programme. Since screening began, through reducing diagnostic delay and speeding-up intervention, around 60 lives have been saved and around 60 cases of long-term disability prevented.

Congenital adrenal hyperplasia (CAH) - CAH is a group of inherited conditions present at birth where the adrenal gland is larger than usual. In CAH the body is missing an enzyme that stimulates the adrenal gland to release cortisol. The 2007 study identified 132 cases (5/100,000 live births). Though the timing of first clinical presentation suggests that almost 70% of babies presenting with salt-wasting crises could benefit from pre-symptomatic detection, screening was not felt appropriate. The study highlighted that test accuracy is poor and showed no improvement in mortality. This view was again endorsed by the NSC in their 2015/16 review which relied heavily on BPSU evidence.

Reye's syndrome - Surveillance of Reye syndrome, a metabolic disorder of unknown origin, predates the establishment of the BPSU and was undertaken by the then Public Health Laboratory Service. By 1986 an association with aspirin had been identified. The need to involve clinicians in reporting the condition led to the formation of the BPSU. The BPSU monitored the impact of the warning to the public not to give children under the age of 12 years aspirin if they had a flu like illness. However though evidence showed a sharp decline in cases of young children, those of teenagers were still being reported. This led the [MHRA to warn against giving aspirin to teenagers](#). Since then virtually no cases have been reported.

Congenital hypothyroidism (CH) - Surveillance of primary CH concluded its surveillance period in June 2012 with follow-up to June 2015. A baby with CH will experience serious learning problems later on in life. Data about CH diagnoses showed laboratories needed to use the same test when screening babies. Data about CH treatment also showed some children no longer need treatment by three years old. This BPSU study found out how many babies in the UK each year have a positive blood spot (heel prick) screening result for CH. The study also helped find out how the screening test could be improved and changes have been made to the screening programme. [Find out more about the findings](#)

Infection and immunisation disease

Guillain–Barré/Miller Fisher syndrome (GB/MFS) - In 2009 following the spread of swine flu from Mexico to Europe, the H1N1 vaccine was introduced in the UK. A previous outbreak in 1976 in the lead-up to a national immunisation programme in the US was discontinued following reports of GB/MFS in some of those who were vaccinated. To monitor the incidence of the conditions following introduction of the vaccine, the BPSU fast tracked surveillance. Very few cases were reported. There appeared to be little association with the H1N1 influenzae vaccination or seasonal flu vaccination. The JCVI have declared that the vaccine is safe for use and it is now in widespread use in the paediatric population.

Public policy

Progressive intellectual and neurological deterioration (PIND) - Following the rise of Bovine spongiform encephalopathy (BSE) in the mid 1990's, there was concern that the prion causing the condition could pass through the food chain to humans. With emerging evidence that this was the case the BPSU was approached by the Department of Health (DoH) to commence surveillance in 1997 for the condition in children. As the diagnosis is one of exclusion a case definition was developed that would identify all children with neurodegenerative conditions. Though the PIND study identified only six cases of variant Creutzfeldt–Jakob disease (vCJD), it has also provided information on over 1,440 cases covering nearly 200 rare neurological disorders. New findings on potential population susceptibilities to the disease (heterozygosity), contamination of blood products, surgical instruments and theoretically via vertical transmission has led the DoH to the view that surveillance should continue with the DoH reliant on the BPSU for monitoring the paediatric population.

Human immunodeficiency virus (HIV) - Since 1986 the BPSU has been the cornerstone of paediatric HIV surveillance in the UK and Ireland. Together with active obstetric HIV surveillance it has comprised a continuous and comprehensive dataset of pregnancies in women with diagnosed HIV infection, their infants and all resident children diagnosed with HIV since 1989. BPSU data has been invaluable in developing recommendations for the monitoring, screening and treatment of HIV, in contributing to modelling studies, and informing long-term planning and commissioning of services for the paediatric population living with HIV.

Haemolytic Uraemic Syndrome (HUS) - HUS was one of the first BPSU studies to be undertaken and which has been repeated on several occasions. In 1986 the first study confirmed definitively the E.coli o157 link with HUS. The third study was fast-tracked following concern over a large outbreak in Europe in the months leading-up to the London Olympics in 2012. The studies, as well as identifying the differing transmission routes, highlighted improved treatment had reduced mortality. Also the identification of a subtype of the condition aHUS led to the establishment of a European cohort. Following recruitment into clinical trials, a new successful but expensive treatment (Eculizumab) was developed and approved.

Congenital rubella - The study has tracked decline in congenital rubella births, and changing demographics of affected mothers and babies, after vaccine strategy changed from selective to mass immunisation. Evidence has been used to measure impact of changes in policy and vaccine uptake, and to inform antenatal screening policy on rubella susceptibility screening, leading to cessation of rubella susceptibility antenatal screening in April 2016. The NSC is now reliant on the BPSU to continue to produce the evidence for considering reviewing the screening policy going forward.

Clinical practice and health management

Biliary Atresia - Evidence from the BPSU survey concluded that patient outcome was improved significantly where surgeons were experienced in performing the Kasai operation. This led to a change in the national service provision of the procedure with the concentration of biliary atresia surgery and management in three national centres. The resultant development of improved guidelines also informed the concentration of services in other European countries eg. Scandinavia and France. 15 years on those outcomes for those

receiving the operation are still being monitored and the overall survival rate has improved from 82% to 94%.

Sudden unexpected postnatal collapse - At the start of the study there was uncertainty of the extent of sudden severe postnatal collapse. Findings indicated that many neonatal units had experienced this problem without any clear guidelines on investigation, management and legal matters. The study findings informed the development of national published guidelines. The findings were also incorporated into the international 'UNICEF Baby Friendly Hospital' guidelines. The evidence from the study has been presented widely nationally and internationally. Also, there have been requests from multiple centres throughout the UK and across the rest of world for further information and for permission to use published materials and in-house resources to help educate staff and improve patient safety.

Severe visual impairment and blindness/congenital cataracts - The BPSU has been working in collaboration with the British Ophthalmology Surveillance Unit to survey rare eye conditions in children. Such joint epidemiological research into childhood visual impairment and blindness has transformed our understanding of the population characteristics and burden, outcomes and impact of childhood visual impairment and blindness in the UK. Changes have resulted in changes to the UK Child Screening and Surveillance Programme, the management of specific conditions, and the assessment and planning of services.

Hyperbilirubinaemia - The BPSU study was quoted by the Chief Medical Officer in their 2004 report. The study led to the development of a National Institute of Clinical Effectiveness (NICE) guideline for investigation and management of neonatal jaundice. The guidelines referenced the BPSU study as an evidence update for the 2010 guidelines.

The NSC considered whether screening for kernicterus was justified by the available evidence. They decided not to recommend this screening, a recommendation in keeping with the literature review findings of the Jaundice Guideline Development Group.

Research and education

Publications - The work of the BPSU and the studies it has facilitated have made a major contribution to research knowledge. Over 250 papers have been published in peer review journals, and a similar number of abstract and conference presentations have been made.

Replication - Other specialty units have been developed in the UK, most notably the British Ophthalmological Surveillance Unit, UK Obstetric Surveillance System, and the Child and Adolescent Psychiatry Surveillance System; all of whom the BPSU has collaborated with. Internationally there are 12 paediatric surveillance units, who in 1996 formed into the International Network of Paediatric Surveillance Units (INOPSU). This has led to research teams sharing research protocols allowing for data comparison.

Expanding NHS research capacity - The BPSU has supported the Sir Peter Tizard Bursary. To date, 14 young doctors have undertaken surveillance projects in order to enhance their understanding of rare disease research. In many cases this has led to further research work.

Conferences - The BPSU has held several workshops and conferences. It has organised sessions within international paediatric conferences.

E-learning - The BPSU has been working with patient support groups to develop e-learning packages on neonatal meningitis and [Kawasaki disease](#).

Information, engagement and advocacy

Patient engagement - The BPSU has been at the forefront when it comes to engaging with lay and patient groups. We have advised and informed researchers on the importance of engaging with patient groups. We have produced literature and held workshops to support active engagement of patients and carers in research proposals. We are now seeing a greater awareness amongst researchers of the importance of active public patient engagement.

The BPSU has been asked to sit on several patient advocacy groups and is represented on [Rare Disease UK's](#) management board and the [Cambridge Rare Disease Network](#), the impact of which can be seen by the willingness of these groups to engage with the paediatric clinical research community.

Advocacy - The BPSU contributed to the [Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy](#) document. This was submitted to the DoH and used as a template for the development of the [UK Strategy for Rare Diseases](#) report for which the BPSU was asked to write a section.

The Chief Medical Officer on more than one occasion has approached the BPSU to contribute to their end-of-year report. Most notably in 2013/14:

We lack nationally collated current data on the present extent of mental health problems and service provision. The last national community survey is a decade old, and both national surveys excluded children under five years old. Prospective surveillance can provide policy and practice relevant data on rare conditions and events that collectively can be costly and difficult to manage, but such surveillance struggles for funding. The Child and Adolescent Psychiatry Surveillance System and the British Paediatric Surveillance Unit use monthly cards to collect data from consultant child and adolescent psychiatrists/paediatricians, sometimes jointly, about a range of rare disorders (early-onset bipolar disorder), conditions (conversion disorder) and events; a study on the cost effectiveness of different types of services for young people with anorexia will commence data collection in the autumn.

Davies, S.C. "Annual Report of the Chief Medical Officer 2013, Public Mental Health Priorities: Investing in the Evidence" London: Department of Health (2014), p.100