

Publishable Executive Summary

Title:

EpiGenChlamydia Consortium

Subtitle:

Chlamydia infections: world leading cause of blindness and tubal pathology

The *EpiGenChlamydia Consortium* is funded by the EU Framework Programme 6 under the Co-ordination Actions in functional genomics research for a period of 2 years (July 2007 –December 2009). The main theme in the *EpiGenChlamydia Consortium* is *Chlamydia trachomatis* (CT) infection which is responsible for both the world leading cause of blindness (trachoma) and the most prevalent sexually transmitted disease which is strongly associated with pelvic inflammatory disease, ectopic pregnancy, and tubal infertility. Prevalences are increasing worldwide with almost 100 million new infections each year.

Twin study-based findings of members of this consortium estimate that there is a 40% genetic predisposition to CT infections (Bailey et al., Drugs of Today, Nov 2009), forming the basis. The overall goal of the *EpiGenChlamydia Consortium* is to accommodate the optimal environment to build and prepare a consortium to reliably determine the genetic predisposition to CT infection, to develop diagnostic tools that can determine individual's predisposition to infection and the risk to develop late complications, and to enhance knowledge of the CT – host interaction, in order to allow the development of novel tools for the detection and treatment of and vaccine development for CT infections.

Meeting schedule:

The Kick-off meeting was held 12th December 2007 and the final meeting was held 15-16 November 2009. In between 2 other meetings were organized and sub-meetings were held, amongst other during the “6th Meeting of the European Society of Chlamydia Research” in Aarhus (Denmark) July 2008. In addition, at the Aarhus meeting we organized an open session entitled “*Immunogenetics of Chlamydia Infection*”, with all international experts in the field.

Objectives and deliverables:

- (1) We provided state-of-the-art reports on the epidemiology of both ocular and sexually transmitted CT infections
- (2) We defined and provided an approach based on the host-pathogen interaction for large-scale genetic typing
- (3) We generate(d) a ethically and ethnic validated and integrated large biobank (in part physically and in part virtually with over 2.2 million samples) and an efficient data warehouse
- (4) We have disseminated our consortium activities within and outside Europe
- (5) We are currently defining the EpiGenChlamydia consortium's future possibilities and have preliminary lobbied a call in the FP7 Health 2010-2011 entitled “*Large-scale genomics approaches to identify host determinants of infectious diseases*”.

Future perspectives and Directions

The aims and work of the EpiGenChlamydia Consortium highly contributes to Public Health. The results of the EpiGenChlamydia Consortium has been chosen by the international Network GRaPHint (Genome-based Research and Population Health) as a best practice example for the development of innovative tools for diagnosis and treatment of Host Genetic Determinants of Infectious Diseases (HGDoID) for the benefit of population health. In close collaboration with the Public Health Agency Canada (PHAC), the Centers for Disease Control and Prevention in the US (CDC) and the European Centre for Disease Prevention and Control (ECDC) a high-level meeting will be held in the end of 2010 in Maastricht, the Netherlands, to increase political awareness about the relevance of this topic for Public Health.

To timely translate the findings of the EpiGenChlamydia Consortium into health policy and practice, the Consortium has been extended by involving the Public Health Genomics European Network (PHGEN). PHGEN is a cornerstone in the development of Public Health Genomics in Europe. It is coordinated from the European Centre of Public Health Genomics (ECPHG) in Maastricht and funded by the General Directorate for Health and Consumer Protection (DG SANCO) under the Health Programme. Within the next three years PHGEN will produce the first edition of "European Best Practice Guidelines for Quality Assurance, Provision and Use of Genome-based Information and Technologies". The guidelines assist Member States, Applicant and EFTA-EEA Countries with evidence-based guidance on timely and responsible integration of genome-based information and technologies into healthcare systems for the benefit of population health.

Keywords:

Chlamydia trachomatis infections, sexually transmitted infections and ocular infection, genetic epidemiology, SNP-Chip, ethnic defined cohorts, host factors, bacterial factors, environmental factors, data warehouse development, biobank development, public health relevance

Project Information:

Acronym: *EpiGenChlamydia Consortium*
Full length title: Contribution of molecular epidemiology and host-pathogen genomics to understand *Chlamydia trachomatis* disease
Contract Number: 037637
Duration: 2 years: 1 July 2007 – 31 December 2009
(half extension request in progress)
Framework Programme: FP6-2005-LIFESCIHEALTH-6 LSH-2005-1.1.0-1
Size: 20 Institutes & Universities including two in Africa and two in the US

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Figure 1 text

Pert Diagram showing the Interdependencies of the EpiGenChlamydia Work-packages (WPs)

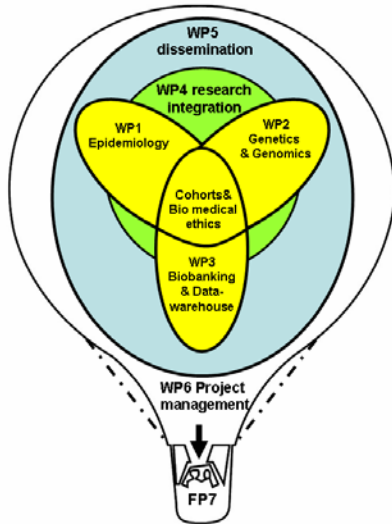


Figure 2 text

Mature inclusion of *C. trachomatis*. The cell cytoplasm is packed with small, dark-staining, chlamydial elementary bodies. The host cell nucleus has been pushed into the left-hand corner of the cell. A cell like this will eventually lyse to release over 10,000 infectious chlamydial elementary bodies which will infect many new cells. Electron micrograph by M. E. Ward.

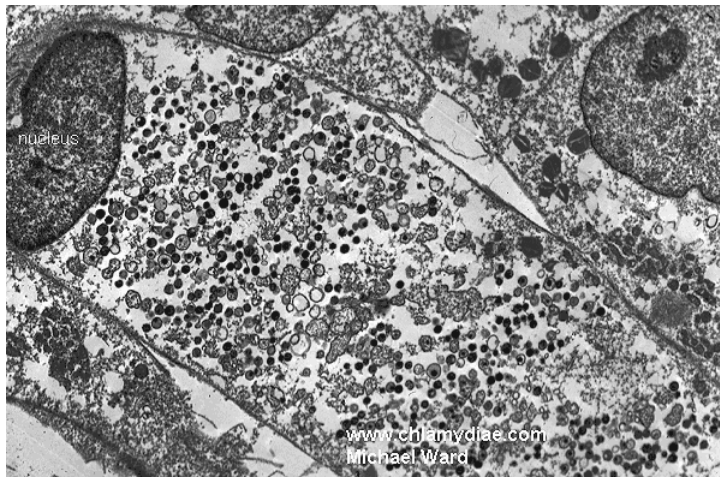
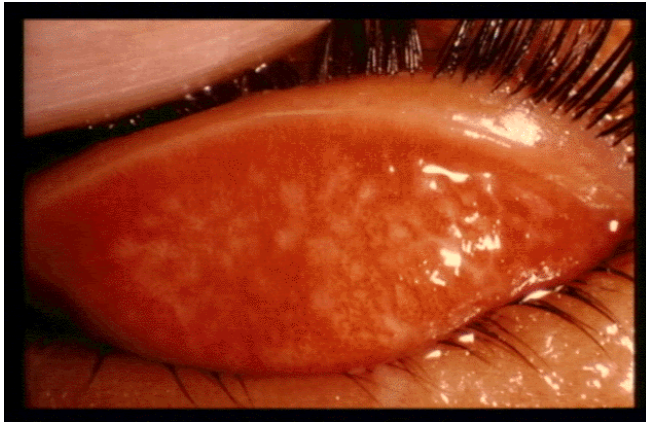


Figure 3 text

Intense trachomatous inflammation. The tarsal conjunctiva appears red, rough and thickened, obscuring more than half of the normal, deep, tarsal vessels. There are numerous follicles which are partially covered by the thickened conjunctiva. The more severe inflammation might give rise to the blinding sequelae of trachoma. Figure kindly provided by the World Health Organisation.



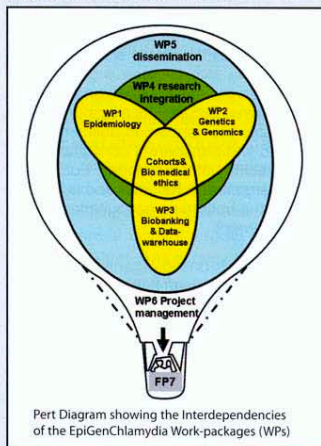
Publishable Executive Summary is based and updated on a publication of an one page dissemination (see below) in the Parliament Magazine 21 September 2009, which was amongst others disseminated at the Gastein European Health Forum Meeting in September 2009.



EpiGenChlamydia Consortium

Chlamydia infections: world leading cause of blindness and tubal pathology

The *EpiGenChlamydia Consortium* is funded by the EU Framework Programme 6 under the Co-ordination Actions in functional genomics research for a period of 30 months (July 2007 – December 2009). The main theme in the *EpiGenChlamydia Consortium* is *Chlamydia trachomatis* (CT) infection which is responsible for both the world leading cause of blindness (trachoma) and the most prevalent sexually transmitted disease which is strongly associated with pelvic inflammatory disease, ectopic pregnancy, and tubal infertility. Prevalences are increasing worldwide with almost 100 million new infections each year.



Twin study-based findings of members of this consortium estimate that there is a 40% genetic predisposition to CT infections (Bailey *et al.*, *Drugs of Today*, Nov 2009). The overall goal of the *EpiGenChlamydia Consortium* is to accommodate the optimal environment to build and prepare a consortium to reliably determine the genetic predisposition to CT infection, to develop diagnostic tools that can determine individual's predisposition to infection and the risk to develop late complications.

Meetings organised by the consortium

The Kick-off meeting was held 12th December 2007 and the final meeting will be held 15-16 November 2009. In between 2 other meetings were organized and sub-meetings were held, amongst other during the "6th Meeting of the European Society of Chlamydia Research" in Aarhus (Denmark) July 2008. In addition, at the Aarhus meeting we organized an open session entitled "Immunogenetics of Chlamydia Infection".

Preliminary results:

- (1) We provided state-of-the-art reports on the epidemiology of both ocular and sexually transmitted CT infections
- (2) We defined and provided an approach based on the host-pathogen interaction for large-scale genetic typing
- (3) We have generated an ethically and ethnic validated and integrated large biobank and an efficient data warehouse
- (4) We have disseminated our consortium activities within and outside Europe
- (5) We are currently defining the EpiGenChlamydia consortium's future funding possibilities.

Future perspectives and Directions

The aims and work of the EpiGenChlamydia Consortium highly contributes to Public Health and we started collaborating with the international Network GRaPH-Int (Genome-based Research and Population Health) on Host Genetic Determinants of Infectious Diseases (HGDoID) for the benefit of population health. In close collaboration with the Public Health Agency Canada (PHAC), the Centers for Disease Control and Prevention in the US (CDC) and the European Centre for Disease Prevention and Control (ECDC) a high-level meeting on HGDoID will be held in the end of 2010 in Maastricht, the Netherlands, to increase political awareness about the relevance of this topic for Public Health.

To timely translate the findings of the EpiGenChlamydia Consortium into health policy and practice, the Consortium is collaborating with the Public Health Genomics European Network (PHGEN) which coordinated from the European Centre of Public Health Genomics (ECPHG)

in Maastricht (Prof. Angela Brand) and funded DG SANCO under the Health Programme. ECPHG develops guidelines to assist Member States, Applicants and EFTA-EEA Countries with evidence-based guidance on timely and responsible integration of genome-based information and technologies into healthcare systems for the benefit of population health. Currently, major efforts of the EpiGenChlamydia

Project Information:

Full length title: Contribution of molecular epidemiology and host-pathogen genomics to understand Chlamydia trachomatis disease
Contract Number: 037637 (in FP6 Health)
Duration: 2.5 years: 1 July 2007 – 31 December 2009
Size: 20 Institutes & Universities including two in Africa and two in the US

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Consortium are directed to obtain funding to use the validated biobank and dataware house for large scale genetic typing to obtain genetic markers to translate in Public Health to combat the severe sequelae of *Chlamydia trachomatis* infections.



A human cell like this will eventually lyse to release over 10,000 infectious chlamydial elementary bodies which will infect many new cells. Electron micrograph by M. E. Ward.