

COST Action Final Achievement Report (11/05/2015 to 10/05/2019)

BM1407: Translational research in primary ciliary dyskinesia - bench, bedside, and population perspectives (BEAT-PCD)

The Action was approved by the Committee of Senior Officials (CSO) on 13-11-2014 and has the MoU reference COST 085/14.

This report was submitted on 06-06-2019 by the Action Chair on behalf of the Management Committee in fulfilment of the requirements of the rules for COST Action Management, Monitoring and Final Assessment.

Action leadership and participants

Leadership positions

| Position | Name | Contact details | Country* |
|----------|-----------------|-------------------------------------|----------------|
| Chair | Prof Jane Lucas | jucas1@soton.ac.uk +442381206160 | United Kingdom |

| Position | Name | Contact details | Country* |
|------------|---------------------|---|-------------|
| Vice Chair | Prof Claudia Kuehni | claudia.kuehni@ispm.unibe.ch +410316313507 | Switzerland |

Working groups

| # | WG Title | # of participants | WG Leader | Country* |
|---|-----------------|-------------------|---|----------------|
| 1 | Basic science | 114 | Dr Dominic Norris d.norris@har.mrc.ac.uk | United Kingdom |
| 2 | Epidemiology | 66 | Prof Claudia Kuehni claudia.kuehni@ispm.unibe.ch | Switzerland |
| 3 | Clinical care | 137 | Dr Kim G Nielsen kgn@dadlnet.dk | Denmark |
| 4 | Clinical trials | 105 | Prof Philipp Latzin philipp.latzin@insel.ch | Switzerland |

Other key leadership positions

| Position | Name | Contact details | Country* |
|-------------------------------|---------------------|--------------------------------|----------------|
| STSM Coordinator | Prof Claire Hogg | c.hogg@rbht.nhs.uk | United Kingdom |
| Science Communication Manager | Dr Myrofora Goutaki | myrofora.goutaki@ispm.unibe.ch | Switzerland |
| GH Scientific Representative | Prof Jane Lucas | jucas1@soton.ac.uk | United Kingdom |

* The country displayed is:

- for the Action Chair, the country that nominated that person to the Management Committee before they were elected Action Chair;
- for the Vice Chair the country that nominated the person as a Management Committee Member,
- for all other leadership positions, if the person is a MC Member the country displayed is the country of nomination, otherwise it is the country of the person's primary work affiliation.

Participants

COST members having accepted the MoU

| | | | | | | | | | |
|-----------|------------|-----------|------------|-----------|------------|-----------|------------|-----------|------------|
| AT | 15/01/2015 | BE | 20/01/2015 | CY | 04/12/2014 | CZ | 14/05/2015 | DK | 05/12/2014 |
| FI | 11/05/2016 | FR | 12/12/2014 | DE | 28/11/2014 | EL | 04/03/2015 | IE | 08/04/2015 |
| IL | 03/12/2014 | IT | 09/02/2015 | NL | 12/03/2015 | NO | 02/02/2015 | PL | 02/12/2014 |
| PT | 02/02/2015 | RS | 17/06/2015 | ES | 10/12/2014 | SE | 06/07/2015 | CH | 16/01/2015 |
| TR | 25/05/2015 | UK | 18/11/2014 | | | | | | |

Other participants

| Institution Name | Country |
|---|---------------|
| Makassed Hospital | Palestine** |
| The Hospital for Sick Children, University of Toronto | Canada |
| The Ohio State University | United States |
| University of Alberta | Canada |
| University of North Carolina | United States |
| Clínica Santa Maria | Chile |

Summary

Main aim/ objective

The main objective of the Action is to create a network of multidisciplinary researchers. The network will promote research from basic science to clinical care, with the ultimate goal to develop treatments that lead to improvements in long-term outcome of patients with PCD.

The Action addressed this as described below

'Better Experimental Approaches to Treat Primary Ciliary Dyskinesia' (BEAT-PCD; COST Action BM1407) comprises a network of scientists and clinicians coordinating research from basic science through to clinical care with the intention of developing treatments and diagnostics that lead to improved long-term outcomes for patients. We united a multidisciplinary network of over 250 participants from 22 COST States and four additional countries. Our collaborations have delivered major advances in clinical care, in particular the first evidence-based guidelines for the diagnosis of PCD, the commissioning of the ERN-LUNG PCD network, and a number of consensus statements. Step changes have also been made through our research collaborations, for example an international PCD cohort (iPCD) of over >3000 patients providing infrastructure for epidemiological studies, a PCD registry to support future clinical trials, the first multinational clinical trial of a treatment for PCD has presented positive results, and we have developed, validated and translated quality of life tools (QOL-PCD) for use as outcome measures in trials.

BEAT-PCD activities are coordinated through four integrated workgroups: basic science, epidemiology, clinical care and clinical trials (outcome measures). Workshops at BEAT-PCD conferences identified and prioritised research projects which are being conducted through the workgroups, to provide data to support H2020 grant applications. For example, the scientists have undertaken a review of pre-clinical models, and have standardised approaches for diagnostic testing. BEAT-PCD identified urgent topics, and have delivered guidelines for clinical management, e.g. prevention of cross-infection, analysing and reporting ciliary ultrastructure and defining respiratory exacerbations.

The active Training School, promotes excellent education, training and career development to its 100 members. Interaction between senior academics, clinicians, students and post-doctoral fellows and invited speakers from academia and industry at conferences, workshops and Training Schools provides career development opportunities. A number of our network have been successfully awarded PhDs, and continue to advance the PCD research arena as postdoctoral scientists. The Training School has included four meetings comprising state-of-the-art lectures from senior academics and clinicians, oral presentations from TS members, poster sessions and educational workshops. Early career researchers/ clinicians are encouraged and supported in management and administrative roles within the Action e.g. management and steering committee, writing reports, newsletters and manuscripts, chairing sessions, oral presentations, and communications.

Four BEAT-PCD conferences and associated meetings of the workgroups have provided a focus for our activities. In addition to experts in PCD, our speakers have included scientists from different disease backgrounds who use cutting edge methods which might be pertinent to PCD research e.g. an expert in super-resolution imaging which might be translated into PCD research, and methodologists seeking statistical approaches for conducting clinical trials in rare diseases.

Outputs from our network include >40 manuscripts in peer reviewed journals and a number of symposium presentations at international meetings.

Action website

<http://www.beatpcd.org/>

Achievement of MoU objectives, deliverables and additional outputs/ achievements

MoU objectives

The Action reported the following achievement of its specific objectives.

| MoU objective | Level of achievement | Further information (hyperlink or other) |
|--|----------------------|--|
| to strengthen the infrastructure for research on PCD | 76 - 100% | <p>We have created a large and active multidisciplinary network; there is excellent collaborations between and within work groups as evidenced by authorship of publications, the cross-discipline BEAT-PCD projects and participation in activities during conferences and training schools (TS).</p> <p>The network has continued to grow throughout the four years.</p> <p>December 2015 (inaugural conference) 162 people had registered as MC members, WG members, trainees or MC observers. Demonstrating the multidisciplinary nature of the network the work group membership was as follows: basic science 73; epidemiology 53; clinical care 112; clinical trials and outcomes 69 (individuals can participate in ≥ 1 WG).</p> <p>April 2016 (end of period 1) 196 people had registered. WG membership was: basic science 100; epidemiology 61; clinical care 112; clinical trials and outcomes 88.</p> <p>April 2017 (end of period 2) 257 people had registered. WG membership was: basic science 114; epidemiology 66; clinical care 137; clinical trials and outcomes 105.</p> <p>April 2018 (end of period 3) 264 people had registered. WG membership was: basic science 118; epidemiology 74; clinical care 138; clinical trials and outcomes 112.</p> <p>http://www.beatpcd.org/the-network/</p> <p>http://www.beatpcd.org/publications/</p> <p>http://www.beatpcd.org/news-and-events/</p> <p>Multidisciplinary networking has led to a strengthened infrastructure for research, building on previous consortia and networks which includes:</p> <ol style="list-style-type: none"> 1. Further developed European Registry of patients with PCD for inclusion in trials (started during FP7 BESTCILIA): https://erj.ersjournals.com/content/47/3/849.long 2. Increased numbers of collaborating centres and patients contributing to the International PCD Cohort (iPCD) for epidemiological research (started during FP7 BESTCILIA): https://erj.ersjournals.com/content/49/1/1601181.long 3. A Guideline for the diagnosis of PCD, ensuring all research recruits are diagnosed in a standardised manner (with European Respiratory Society PCD Task Force): https://erj.ersjournals.com/content/49/1/1601090.long 4. The multidisciplinary nature of the network has been important for ensuring scientists and clinicians understand each other's perspectives, and that we have been able to support each other's research. |
| building on and unifying the expert | 76 - 100% | <p>Since year one we have held a number of networking opportunities for experts in the field, developing a common understanding of complex issues through</p> |

networks of
researchers in PCD

multidisciplinary meetings and workshops. These have led to a number of guidelines, consensus statements and research projects:

2015-16 The key activities were the inaugural conference (Southampton, UK), Training School (Paris, France) and a meeting during European Respiratory-ERS Conference (Amsterdam, Netherlands). All meetings successfully integrated scientists, clinicians, patient representatives and industry. At each meeting new projects and collaborations have been planned and implemented through a series of workshops, all working towards the COST Action's mission. The ERS meeting was held jointly with FP7 BESTCILIA.

<https://bmcpoc.biomedcentral.com/articles/10.1186/s12919-016-0067-0>

2016-17: Key activities were a joint conference and training school, Valencia April 2017, with 119 participants. The joint meeting was attended by 108 delegates from 24 countries including 58 trainees. The meeting included key note talks, short oral presentations, STSM presentations, a number of scientific and clinical workshops for all delegates, three workshops of experts to develop guidelines essential for future research and clinical care (definition of exacerbations, segregation of patients to avoid cross-infection, analysis and reporting of electron microscopy), and a poster session. Workgroups met during the conference to plan and progress projects.

<https://bmcpoc.biomedcentral.com/articles/10.1186/s12919-018-0098-9>

The PCD Core group of the recently approved European Reference Network on rare lung diseases (ERN-LUNG) held a session during the BEATPCD conference to which interested parties who are not yet members were invited.

<https://ern-lung.eu/>

https://ec.europa.eu/health/sites/health/files/ern/docs/2017_brochure_en.pdf

https://epar.iplep.upmc.fr/site_respirare/images/Docs/ern/ern_lung_publi.pdf

A very successful meeting was held during the ERS Congress, London September 2016, attended by >80 senior academics and clinicians, and early year researchers/ clinicians.

2017-18

BEATPCD met during the ERS Congress in Milan September 2017, and had presentations about the state-of-the-art diagnostics and therapeutics.

The third BEAT-PCD conference and training school were held jointly in February 2018 in Lisbon (121 participants), Portugal. Presentations and workshops focused on advancing the knowledge and skills relating to PCD in: basic science, epidemiology, diagnostic testing, clinical management and clinical trials. The multidisciplinary conference provided an interactive platform for exchanging ideas through a program of lectures, poster presentations, breakout sessions and workshops. Three work groups met to plan consensus statements. Progress with BEAT-PCD projects was shared and new collaborations were fostered.

<https://bmcpoc.biomedcentral.com/articles/10.1186/s12919-018-0161-6>

BEAT-PCD met during the ERS Congress in Milan September 2017, and had presentations about the state-of-the-art diagnostics and therapeutics.

2018-19 BEAT-PCD met during the ERS Congress in Paris September 2018.

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| | | <p>There was a series of state-of-the art talks. Representatives of the European and American Diagnostic Guidelines discussed the differences with an aim of resolving confusion caused by two guidelines. A joint European-North American statement is being prepared as a consequence of the meeting.</p> <p>The final conference and training school were held in Poznan, Poland, welcoming 145 participants. The successes of the network were discussed including consensus statements, guidelines and infrastructure. Workshops further developed our projects, and we planned for ongoing activities.</p> |
| providing targeted training for ESRs in Training Schools and on Short-Term Scientific Missions, in the clinical and basic science aspects of PCD | 76 - 100% | <p>The network of Early Career Researchers has been particularly active, ensuring the sustainability of the clinical and research programme for decades to come. Four training schools have been completed, three of them alongside the annual BEATPCD conference. The training schools have maintained a programme of lectures, workshops, oral presentations and poster sessions. As well as specific training in PCD, the network has included ECRs in all aspects of our activities, providing opportunities for gaining generic skills such as organising conferences and workshops, media and communications, project managing international research and chairing plenary sessions. Feedback has been excellent.</p> <p>ECRs have initiated and led a networking programme, meeting annually and communicating through our project management tool (Basecamp) and at the Training School.</p> <p>STSM uptake has been positive, providing opportunities for ECRs to gain new methods, and to start building a collaboration network independent of their seniors.</p> |
| building on standardized prospective data collection, developing infrastructure for data and sample sharing | 76 - 100% | <ol style="list-style-type: none"> 1. A multicentre project has developed a standardised PCD proforma for clinical follow- up and prospective data collection. The proforma is now undergoing pilot testing in clinics. The project has involved 40 members from 15 countries. 2. We have templates for data sharing agreements for collaborative projects (eg. iPCD cohort, PROVALF-PCD and European PCD Registry). 3. BEAT-PCD has enabled face-to-face communications and discussions for data which has facilitated sharing between international and national datasets, registries and studies (iPCD cohort, European PCD registry, RaDiCo etc). Exchange of eCRFs, identification of data sharing problems and research for solutions. 4. An expert Working Group has developed and validated a protocol for diagnostic analysis by transmission electron microscopy. The international use of standardised nomenclature and reporting is an important step, and projects are ongoing for other diagnostic tests (Immunofluorescence labelling, High-Speed Video Microscopy). "International consensus guideline for reporting transmission electron microscopy results in the diagnosis of PCD" have been submitted for peer review publication. 5. A BEAT-PCD expert clinical Workin Group developed a consensus definition for Pulmonary Exacerbations for use as an outcome measure in clinical trials. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6355979/ |
| to promote and integrate basic research on PCD to underpin translational research with the ultimate aim to develop novel therapies | 76 - 100% | <p>We have promoted basic research through the annual BEAT-PCD and training school meetings. This has included research talks from invited speakers and BEAT-PCD members. The sharing of ideas and information, at these meetings and beyond these meetings, has both underpinned the research and established new networks and collaborations. Through STSMs, these relationships have been further cemented.</p> <p>Ongoing discussion of gene therapy approaches in Valencia, Lisbon and Poznan, between basic science and applied clinical groups has been exploring the possibilities of novel therapies</p> |

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| <p>to provide representative data on the epidemiology of PCD e.g. long-term prognosis and how the latter is influenced by environmental exposures and different management strategies</p> | <p>76 - 100%</p> | <p>Throughout the four years the participation of countries and centres to the iPCD cohort has grown (currently containing data of >3800 patients from 22 countries) and there was enrichment of the available datasets. Similarly participation in the European PCD Registry has grown. A PCD project investigating the longitudinal change in lung function (PROVALF) is prospectively collecting project specific data, which is also contributing the iPCD and the Registry where data agreements are in place.</p> <p>Data from these epidemiological projects have been presented at a number of international meetings, and a number of publications have been published in high impact journals, including:</p> <p>Description of the European Respiratory Registry: https://erj.ersjournals.com/content/47/3/849.long</p> <p>Description of the iPCD Cohort: https://erj.ersjournals.com/content/49/1/1601181.long</p> <p>Growth and Nutritional status in the iPCD Cohort: https://erj.ersjournals.com/content/50/6/1701659.long</p> <p>Lung Function in the iPCD Cohort: https://erj.ersjournals.com/content/52/2/1801040.long</p> |
| <p>to improve clinical care for patients with PCD</p> | <p>76 - 100%</p> | <p>The BEAT-PCD network has provided an excellent opportunity for clinicians from Europe and beyond to meet and share knowledge. For a rare disease, this international coordination is essential to improve diagnosis and treatments. We have had patient representatives playing key roles at our conferences, and also in expert consensus statements. This has ensured we maintain a patient-centred approach to improving care. Advances over the past four year are highly significant in quality and quantity.</p> <p>Examples include:</p> <ul style="list-style-type: none"> • The first international evidence based guideline for the diagnosis of PCD (with European Respiratory Society): https://erj.ersjournals.com/content/49/1/1601090.long • An expert statement on the management of children with PCD: http://www.tandfonline.com/eprint/x9aFtKuTFuiGxGHmP2K/full • Through a series of workshops at BEAT-PCD conferences, the Work Group has conducted an international survey about the treatment strategies and management of Pseudomonas infection in different PCD centres . This survey is now feeding in to an ongoing project to develop an international consensus statement concerning infection control and treatment. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6302970/ • We are scoping the service delivery for PCD across Europe. A series of semi- structured interviews have been conducted with clinicians. A survey of clinicians is now in progress. This project is being led by an ECR as part of her PhD project, and will be submitted for peer reviewed publication within the next year. • An international network of physiotherapists who specialize in PCD has formed as a direct result of BEAT-PCD. • A consensus statement for using TEM as a diagnostic tool has been developed by experts from 18 centres in 14 countries. The reporting and nomenclature has been validated, and submitted for peer review publication. • A chapter has been published in an educational book regarding management of PCD: Kuehni CE, Goutaki M, Rubbo B, Lucas JS. Management of primary ciliary dyskinesia: current practice and future perspectives. In: Chalmers JD, Polverino E, Aliberti S, eds. Bronchiectasis (ERS Monograph). Sheffield, European Respiratory Society, 2018; pp. 282–299 [https://doi.org/10.1183/2312508X.10016717]. |

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| <p>to improve the evidence base for treatment of PCD, by planning the conduct of clinical trials</p> | <p>76 - 100%</p> | <p>1. The first international clinical trial in PCD, which started during FP7 BESTCILIA, has continued within the network of BEAT-PCD. BEAT-PCD has held workshops and talks to learn from the trial of azithromycin to prevent pulmonary exacerbations. The study protocol is published, and the first results of the study were presented at the BEAT-PCD conference in Poznan, March 2019. https://bmcpulmed.biomedcentral.com/articles/10.1186/s12890-016-0261-x</p> <p>2. We have conducted systematic reviews to better understand outcome measures which might be used for clinical trials. The systematic reviews are complete, results have been disseminated at international conferences, the first manuscript is published and two more publications are in preparation for publication. https://openres.ersjournals.com/content/5/2/00231-2018</p> |
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Deliverables

The Action reported the following deliverables:

| Deliverable | Timing of deliverable | Further information (hyperlink or other) |
|--|--|---|
| Manuscript evaluating the currently available pre-clinical models for PCD, and analysis of what is still needed. | Not delivered, but foreseen within 2 years | |
| Manuscript describing the iPCD cohort | Delivered | https://erj.ersjournals.com/content/49/1/1601181.long |
| At least 2 manuscripts analysing cross sectional data from iPCD (e.g. lung function, growth) | Delivered | https://erj.ersjournals.com/content/52/2/1801040.long |
| At least 1 manuscript analysing longitudinal data from iPCD cohort (eg. lung function) | Delivered | https://erj.ersjournals.com/content/48/suppl_60/PA3127 |
| A proforma for standardised collection of clinical research data. | Not delivered, but foreseen within 2 years | |
| Systematic review looking at outcome measures for PCD clinical trials | Delivered | https://openres.ersjournals.com/content/5/2/00231-2018 |
| Manuscript discussing new outcome measures in PCD | Not delivered, but foreseen within 2 years | |
| Publication of validation studies of QOL-PCD | Delivered | http://thorax.bmj.com/content/early/2017/02/28/thoraxjnl-2016-209356.long |
| Translation of QOL-PCD into at least 7 languages | Delivered | https://erj.ersjournals.com/content/48/suppl_60/PA4161 |
| Evidence based guideline for the diagnosis of PCD | Delivered | https://www.ncbi.nlm.nih.gov/pubmed/27836958 |
| Consensus statement for defining respiratory exacerbations for clinical trials | Delivered | https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6355979/ |
| Consensus statement for preventing cross infections in patients with PCD | Not delivered, but foreseen within 2 years | |
| Consensus statement for TEM as a diagnostic tool | Not delivered, but foreseen within 2 years | |
| Proceedings report from Inaugural Conference | Delivered | https://bmcproc.biomedcentral.com/articles/10.1186/s12919-016-0067-0 |
| Proceedings report from 2nd BEATPCD conference | Delivered | https://bmcproc.biomedcentral.com/articles/10.1186/s12919-018-0098-9 |
| Proceedings document from 3rd BEATPCD conference | Delivered | https://bmcproc.biomedcentral.com/articles/10.1186/s12919-018-0161-6 |
| 24 month report to the COST Action office | Delivered | https://e-services.cost.eu |
| Final Report to the COST Action office | Delivered | https://e-services.cost.eu/action/report/final/achievement/BM1407/deliverables/D_18 |

Additional outputs/ achievements

The following outputs/ achievements also resulted from the Action:

The Action reported 39 publications on the topic of the Action, co-authored by at least two Action participants from two countries participating in the Action, and for which the Action networking was necessary.

Co-authored Action publications - peer-reviewed

1. [doi:10.1183/23120541.00147-2018](https://doi.org/10.1183/23120541.00147-2018)Title

Pulmonary exacerbations in patients with primary ciliary dyskinesia: an expert consensus definition for use in clinical trials

Authors [Jane S. Lucas](#); Florian Gahleitner; Adelina Amorim; Mieke Boon; Philippa Brown; [Carolina Constant](#); Simon Cook; Suzanne Crowley; Damien M.S. Destouches; Ernst Eber; Huda Mussaffi; Eric Haarman; Amanda Harris; Cordula Koerner-Rettberg; [Claudia E. Kuehni](#); [Philipp Latzin](#); Michael R. Loebinger; Natalie Lorent; Bernard Maitre; Antonio Moreno-Galdó; Kim G. Nielsen; Uğur Özçelik; Lue Katrine Drasbæk Philipsen; Petr Pohunek; Eva Polverino; Jessica Rademacher; Phil Robinson; Deborah Snijders; Panayiotis Yiallourous; [Siobhán B. Carr](#)

DOI [doi:10.1183/23120541.00147-2018](https://doi.org/10.1183/23120541.00147-2018)

Type Journal article

Published in ERJ Open Research

Published by European Respiratory Society (ERS)

ISSN [2312-0541](https://doi.org/10.1183/23120541)

Link <https://syndication.highwire.org/content/doi/10.1183/23120541.00147-2018>
2. [doi:10.1016/j.ajhg.2018.10.016](https://doi.org/10.1016/j.ajhg.2018.10.016)Title

Mutations in Outer Dynein Arm Heavy Chain DNAH9 Cause Motile Cilia Defects and Situs Inversus

Authors Mahmoud R. Fassad; Amelia Shoemark; Marie Legendre; Robert A. Hirst; France Koll; Pierrick le Borgne; Bruno Louis; Farheen Daudvohra; Mitali P. Patel; Lucie Thomas; Mellisa Dixon; Thomas Burgoyne; Joseph Hayes; Andrew G.

| | | |
|----|---|--|
| | DOI | doi:10.1016/j.ajhg.2018.10.016 |
| | Type | Journal article |
| | Published in | The American Journal of Human Genetics |
| | Published by | Elsevier BV |
| | ISSN | 0002-9297 |
| | Links | https://api.elsevier.com/content/article/PII:S0002929718303689?httpAccept=text/xml; https://api.elsevier.com/content/article/PII:S0002929718303689?httpAccept=text/plain |
| 3. | doi:10.1016/j.ajhg.2018.10.003 Title | |
| | Authors | Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects Serge Bonnefoy; Christopher M. Watson; Kristin D. Kernohan; Moara Lemos; Sebastian Hutchinson; James A. Poulter; Laura A. Crinnion; Ian Berry; Jennifer Simmonds; Pradeep Vasudevan; Chris O'Callaghan; Robert A. Hirst; Andrew Rutman; Lijia Huang; Taila Hartley; David Grynspan; Eduardo Moya; Chunmei Li; Ian M. Carr; David T. Bonthron; Michel Leroux; Kym M. Boycott; Philippe Bastin; Eamonn G. Sheridan |
| | DOI | doi:10.1016/j.ajhg.2018.10.003 |
| | Type | Journal article |
| | Published in | The American Journal of Human Genetics |
| | Published by | Elsevier BV |
| | ISSN | 0002-9297 |
| | Subjects | Genetics(clinical); Genetics |
| | Links | https://api.elsevier.com/content/article/PII:S0002929718303550?httpAccept=text/xml; https://api.elsevier.com/content/article/PII:S0002929718303550?httpAccept=text/plain |
| 4. | doi:10.1183/13993003.01040-2018 Title | |
| | Authors | Lung function in patients with primary ciliary dyskinesia: an iPCD Cohort study Florian S. Halbeisen; Myrofora |

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|----|---|--|
| | | <p>Goutaki; Ben D. Spycher; Israel Amirav; Laura Behan; Mieke Boon; Claire Hogg; Carmen Casaulta; Suzanne Crowley; Eric G. Haarman; Bulent Karadag; Cordula Koerner-Rettberg; Michael R. Loebinger; Henryk Mazurek; Lucy Morgan; Kim G. Nielsen; Heymut Omran; Francesca Santamaria; Nicolaus Schwerk; Guillaume Thouvenin; Panayiotis Yiallourous; Jane S. Lucas; Philipp Latzin; Claudia E. Kuehni</p> |
| | DOI | doi:10.1183/13993003.01040-2018 |
| | Type | Journal article |
| | Published in | European Respiratory Journal |
| | Published by | European Respiratory Society (ERS) |
| | ISSNs | 0903-1936 ; 1399-3003 |
| | Subject | Pulmonary and Respiratory Medicine |
| | Link | https://syndication.highwire.org/content/doi/10.1183/13993003.01040-2018 |
| 5. | doi:10.1177/1479972318787919 Title | Variation in treatment strategies for the eradication of <i>Pseudomonas aeruginosa</i> in primary ciliary dyskinesia across European centers |
| | Authors | Suzanne Crowley ; Mathias Geldermann Holgersen ; Kim Gjerum Nielsen |
| | DOI | doi:10.1177/1479972318787919 |
| | Type | Journal article |
| | Published in | Chronic Respiratory Disease |
| | Published by | SAGE Publications |
| | ISSNs | 1479-9731 ; 1479-9731 |
| | Subject | Pulmonary and Respiratory Medicine |
| | Links | http://journals.sagepub.com/doi/pdf/10.1177/1479972318787919 ; http://journals.sagepub.com/doi/full-xml/10.1177/1479972318787919 |
| 6. | doi:10.1016/j.ajhg.2018.03.025 Title | Mutations in C11orf70 Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry Due to Defects of Outer and Inner Dynein Arms |
| | Authors | Inga M. Höben; Rim Hjeij; Heike Olbrich; Gerard W. Dougherty; Tabea Nöthe-Menchen; Isabella |

Aprea; Diana Frank; Petra Pennekamp; Bernd Dworniczak; Julia Wallmeier; Johanna Raidt; Kim G. Nielsen; Maria C. Philipsen; Francesca Santamaria; Laura Venditto; Israel Amirav; Huda Mussaffi; Freerk Prenzel; Kaman Wu; Zeineb Bakey; Miriam Schmidts; Niki T. Loges; Heymut Omran

DOI

[doi:10.1016/j.ajhg.2018.03.025](https://doi.org/10.1016/j.ajhg.2018.03.025)

Type

Journal article

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Links

<https://api.elsevier.com/content/article/PII:S0002929718301149?httpAccept=text/xml>;

<https://api.elsevier.com/content/article/PII:S0002929718301149?httpAccept=text/plain>

7. [doi:10.1016/j.ajhg.2018.03.024](https://doi.org/10.1016/j.ajhg.2018.03.024)Title

C11orf70 Mutations Disrupting the Intraflagellar Transport-Dependent Assembly of Multiple Axonemal Dyneins Cause Primary Ciliary Dyskinesia
 Mahmoud R. Fassad; Amelia Shoemark; Pierrick le Borgne; France Koll; Mitali Patel; Mellisa Dixon; Jane Hayward; Charlotte Richardson; Emily Frost; Lucy Jenkins; Thomas Cullup; Eddie M.K. Chung; Michel Lemullois; Anne Aubusson-Fleury; Claire Hogg; David R. Mitchell; Anne-Marie Tassin; Hannah M. Mitchison

Authors

DOI

[doi:10.1016/j.ajhg.2018.03.024](https://doi.org/10.1016/j.ajhg.2018.03.024)

Type

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8. [doi:10.1183/13993003.01659-2017](https://doi.org/10.1183/13993003.01659-2017)Title

Growth and nutritional status, and their association with lung function: a study from the international Primary Ciliary Dyskinesia Cohort

Authors

[Myrofora Goutaki](https://www.elsevier.com/author/Myrofora-Goutaki); Florian S.

Halbeisen; Ben D. Spycher;
 Elisabeth Maurer; Fabiën Belle;
[Israel Amirav](#); Laura Behan;
 Mieke Boon; Siobhan Carr;
 Carmen Casaulta; Annick
 Clement; Suzanne Crowley;
 Sharon Dell; Thomas Ferkol;
 Eric G. Haarman; Bulent
 Karadag; Michael Knowles;
 Cordula Koerner-Rettberg;
 Margaret W. Leigh; Michael R.
 Loebinger; Henryk Mazurek;
 Lucy Morgan; Kim G. Nielsen;
 Maria Phillipsen; Scott D.
 Sagel; Francesca Santamaria;
 Nicolaus Schwerk; Panayiotis
 Yiallourous; [Jane S. Lucas](#);
[Claudia E. Kuehni](#)

DOI [doi:10.1183/13993003.01659-2017](https://doi.org/10.1183/13993003.01659-2017)
 Type Journal article
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 ISSNs [0903-1936](#); [1399-3003](#)
 Subject Pulmonary and Respiratory Medicine
 Link <https://syndication.highwire.org/content/doi/10.1183/13993003.01659-2017>

9. [doi:10.1093/hmg/ddx422](https://doi.org/10.1093/hmg/ddx422)Title

Authors

DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport
 Verity L Hartill; Glenn van de Hoek; Mitali P Patel; Rosie Little; Christopher M Watson; Ian R Berry; Amelia Shoemark; Dina Abdelmottaleb; Emma Parkes; Chiara Bacchelli; Katarzyna Szymanska; Nine V Knoers; Peter J Scambler; Marius Ueffing; Karsten Boldt; Robert Yates; Paul J Winyard; Beryl Adler; Eduardo Moya; Louise Hattingh; Anil Shenoy; Claire Hogg; Eamonn Sheridan; Ronald Roepman; Dominic Norris; Hannah M Mitchison; Rachel H Giles; [Colin A Johnson](#)

DOI [doi:10.1093/hmg/ddx422](https://doi.org/10.1093/hmg/ddx422)
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 ISSNs [0964-6906](#); [1460-2083](#)
 Link <http://academic.oup.com/hmg/article-pdf/27/3/529/24325927/ddx422.pdf>

10. [doi:10.1016/j.chest.2017.06.053](https://doi.org/10.1016/j.chest.2017.06.053)Title

Exploring the Art of Ciliary

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| <p>Authors</p> | <p>Beating Jane S. Lucas; Hazel J. Evans; Eric G. Haarman; Robert A. Hirst; Claire Hogg; Claire L. Jackson; Kim G. Nielsen; Heymut Omran; Jean Francois Papon; Phil Robinson; Amelia Shoemark; Woolf T. Walker doi:10.1016/j.chest.2017.06.053</p> |
| <p>DOI</p> | <p>doi:10.1016/j.chest.2017.06.053</p> |
| <p>Type</p> | <p>Journal article</p> |
| <p>Published in</p> | <p>Chest</p> |
| <p>Published by</p> | <p>Elsevier BV</p> |
| <p>ISSN</p> | <p>0012-3692</p> |
| <p>Links</p> | <p>https://api.elsevier.com/content/article/PII:S0012369217327575?httpAccept=text/xml; https://api.elsevier.com/content/article/PII:S0012369217327575?httpAccept=text/plain</p> |
| <p>11. doi:10.1016/j.rmed.2017.08.028Title</p> | <p>Clinical impact of Pseudomonas aeruginosa colonization in patients with Primary Ciliary Dyskinesia</p> |
| <p>Authors</p> | <p>Malena Cohen-Cymerknoh; Nir Weigert; Alex Gileles-Hillel; Oded Breuer; Natalia Simanovsky; Mieke Boon; Kris De Boeck; Angelo Barbato; Deborah Snijders; Mirella Collura; Ugo Pradal; Hannah Blau; Huda Mussaffi; Mareike Price; Lea Bentur; Michal Gur; Micha Aviram; Elie Picard; Michal Shteinberg; Galit Livnat; Joseph Rivlin; Nurith Hiller; David Shoseyov; Israel Amirav; Eitan Kerem</p> |
| <p>DOI</p> | <p>doi:10.1016/j.rmed.2017.08.028</p> |
| <p>Type</p> | <p>Journal article</p> |
| <p>Published in</p> | <p>Respiratory Medicine</p> |
| <p>Published by</p> | <p>Elsevier BV</p> |
| <p>ISSN</p> | <p>0954-6111</p> |
| <p>Links</p> | <p>https://api.elsevier.com/content/article/PII:S0954611117303062?httpAccept=text/xml; https://api.elsevier.com/content/article/PII:S0954611117303062?httpAccept=text/plain</p> |
| <p>12. doi:10.1183/20734735.008517Title</p> | <p>Diagnosis of primary ciliary dyskinesia: summary of the ERS Task Force report</p> |
| <p>Authors</p> | <p>Claudia E. Kuehni; Jane S. Lucas</p> |
| <p>DOI</p> | <p>doi:10.1183/20734735.008517</p> |
| <p>Type</p> | <p>Journal article</p> |
| <p>Published in</p> | <p>Breathe</p> |
| <p>Published by</p> | <p>European Respiratory Society</p> |

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| | ISSNs | (ERS) 1810-6838 ; 2073-4735 |
| | Subject | Pulmonary and Respiratory Medicine |
| | Link | https://syndication.highwire.org/ content/doi/10.1183/20734735.0085 17 |
| 13. | doi:10.1183/13993003.00612-2017 | Title Primary ciliary dyskinesia ciliated airway cells show increased susceptibility to Haemophilus influenzae biofilm formation |
| | Authors | Woolf T. Walker; Claire L. Jackson; Raymond N. Allan; Samuel A. Collins ; Michael J. Kelso; Ardeshir Rineh; Nageshwar R. Yepuri; Ben Nicholas; Laurie Lau; David Johnston; Peter Lackie; Saul N. Faust; Jane S.A. Lucas ; Luanne Hall-Stoodley |
| | DOI | doi:10.1183/13993003.00612-2 017 |
| | Type | Journal article |
| | Published in | European Respiratory Journal |
| | Published by | European Respiratory Society (ERS) |
| | ISSNs | 0903-1936 ; 1399-3003 |
| | Link | https://syndication.highwire.org/ content/doi/10.1183/13993003. 00612-2017 |
| 14. | doi:10.1080/17476348.2017.1360770 | Title Clinical care of children with primary ciliary dyskinesia |
| | Authors | Jane S. Lucas; Mikkel Christian Alanin; Samuel Collins; Amanda Harris; Helle Krogh Johansen; Kim G Nielsen; Jean Francois Papon; Phil Robinson; Woolf T. Walker |
| | DOI | doi:10.1080/17476348.2017.1 360770 |
| | Type | Journal article |
| | Published in | Expert Review of Respiratory Medicine |
| | Published by | Informa UK Limited |
| | ISSNs | 1747-6348 ; 1747-6356 |
| | Link | https://www.tandfonline.com/d oi/pdf/10.1080/17476348.201 7.1360770 |
| 15. | doi:10.3389/fped.2017.00135 | Title Primary Ciliary Dyskinesia: An Update on Clinical Aspects, Genetics, Diagnosis, and Future Treatment Strategies |
| | Authors | Virginia Mirra; Claudius Werner; |

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| DOI Type Published in Published by ISSN Link | Francesca Santamaria doi:10.3389/fped.2017.00135 Journal article Frontiers in Pediatrics Frontiers Media SA 2296-2360 http://journal.frontiersin.org/article/10.3389/fped.2017.00135/full |
| 16. doi:10.1002/humu.23261 Title Authors DOI Type Published in Published by ISSN Links | Mutation of serine/threonine protein kinase 36 (STK36) causes primary ciliary dyskinesia with a central pair defect Christine Edelbusch; Sandra Cindrić ; Gerard W. Dougherty; Niki T. Loges; Heike Olbrich; Joseph Rivlin; Julia Wallmeier; Petra Pennekamp; Israel Amirav; Heymut Omran doi:10.1002/humu.23261 Journal article Human Mutation Wiley 1059-7794 https://api.wiley.com/onlinelibrary/tdm/v1/articles/10.1002%2Fhumu.23261 ; https://onlinelibrary.wiley.com/doi/full/10.1002/humu.23261 |
| 17. doi:10.1186/s12919-018-0161-6 Title Authors DOI Type Published in Published by ISSN Link | Proceedings of the 3rd BEAT-PCD Conference and 4th PCD Training School Hannah Farley; Bruna Rubbo; Zuzanna Bukowy-Bieryllo; Mahmoud Fassad; Myrofora Goutaki; Katharine Harman; Claire Hogg; Claudia E. Kuehni; Susana Lopes; Kim G. Nielsen; Dominic P. Norris; Ana Reula; Nisreen Rumman; Amelia Shoemark; Hannah Wilkins; Agatha Wisse; Jane S. Lucas; June K. Marthin doi:10.1186/s12919-018-0161-6 Journal article BMC Proceedings Springer Nature 1753-6561 http://link.springer.com/content/pdf/10.1186/s12919-018-0161-6.pdf |
| 18. doi:10.1186/s12919-018-0098-9 Title Authors | Proceedings of the 2nd BEAT-PCD conference and 3rd PCD training school: part 1 Florian Halbeisen; Claire Hogg; Mikkel C. Alanin; Zuzanna Bukowy-Bieryllo; Francisco Dasi; |

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| | | Julie Duncan; Amanda Friend; Myrofora Goutaki; Claire Jackson; Victoria Keenan; Amanda Harris; Robert A. Hirst; Philipp Latzin; Gemma Marsh; Kim Nielsen; Dominic Norris; Daniel Pellicer; Ana Reula; Bruna Rubbo; Nisreen Rumman; Amelia Shoemark; Woolf T. Walker; Claudia E. Kuehni; Jane S. Lucas |
| | DOI | doi:10.1186/s12919-018-0098-9 |
| | Type | Journal article |
| | Published in | BMC Proceedings |
| | Published by | Springer Nature |
| | ISSN | 1753-6561 |
| | Subjects | General Biochemistry, Genetics and Molecular Biology; General Medicine |
| | Link | http://link.springer.com/content/pdf/10.1186/s12919-018-0098-9.pdf |
| 19. | doi:10.1186/s12919-016-0067-0 | Title |
| | Authors | Proceedings of the COST action BM1407 inaugural conference BEAT-PCD: translational research in primary ciliary dyskinesia - bench, bedside, and population perspectives Bruna Rubbo; Laura Behan; Eleonora Dehlink; Myrofora Goutaki; Claire Hogg; Panayiotis Kouis; Claudia E. Kuehni; Philipp Latzin; Kim Nielsen; Dominic Norris; Sylvia Nyilas; Mareike Price; Jane S. Lucas |
| | DOI | doi:10.1186/s12919-016-0067-0 |
| | Type | Journal article |
| | Published in | BMC Proceedings |
| | Published by | Springer Nature |
| | ISSN | 1753-6561 |
| | Links | http://link.springer.com/content/pdf/10.1186/s12919-016-0067-0.pdf ; http://link.springer.com/article/10.1186/s12919-016-0067-0/fulltext.html |
| 20. | doi:10.1513/AnnalsATS.201712-967OC | Title |
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| | DOI | |
| | Type | Journal article |
| | Published in | Annals of the American Thoracic Society |
| | Published by | American Thoracic Society |
| | ISSNs | 2329-6933 ; 2325-6621 |
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| 21. | doi:10.1183/13993003.02514-2016 | Title |
| | | Hypertonic saline in patients with primary ciliary dyskinesia: on the road to evidence-based treatment for a rare lung disease |
| | Authors | Claudia E. Kuehni; Myrofora Goutaki; Helene E. Kobbernagel |
| | DOI | doi:10.1183/13993003.02514-2016 |
| | Type | Journal article |
| | Published in | European Respiratory Journal |
| | Published by | European Respiratory Society (ERS) |
| | ISSNs | 0903-1936 ; 1399-3003 |
| | Subject | Pulmonary and Respiratory Medicine |
| | Link | https://syndication.highwire.org/content/doi/10.1183/13993003.02514-2016 |
| 22. | doi:10.1183/13993003.00466-2016 | Title |
| | | Alternative inert gas washout outcomes in patients with primary ciliary dyskinesia |
| | Authors | Sylvia Nyilas; Anne Schlegtendal; Florian Singer; Myrofora Goutaki; Claudia E. Kuehni; Carmen Casaulta; Philipp Latzin; Cordula Koerner- Rettberg |
| | DOI | doi:10.1183/13993003.00466-2016 |
| | Type | Journal article |
| | Published in | European Respiratory Journal |
| | Published by | European Respiratory Society (ERS) |
| | ISSNs | 0903-1936 ; 1399-3003 |
| | Subject | Pulmonary and Respiratory Medicine |

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| | Link | https://syndication.highwire.org/content/doi/10.1183/13993003.00466-2016 |
| 23. | doi:10.1183/13993003.01181-2016 Title | The international primary ciliary dyskinesia cohort (IPCD Cohort): methods and first results |
| | Authors | Myrofora Goutaki; Elisabeth Maurer; Florian S. Halbeisen; Israel Amirav; Angelo Barbato; Laura Behan; Mieke Boon; Carmen Casaulta; Annick Clement; Suzanne Crowley; Eric Haarman; Claire Hogg; Bulent Karadag; Cordula Koerner-Rettberg; Margaret W. Leigh; Michael R. Loebinger; Henryk Mazurek; Lucy Morgan; Kim G. Nielsen; Heymut Omran; Nicolaus Schwerk; Sergio Scigliano; Claudius Werner; Panayiotis Yiallourous; Zorica Zivkovic; Jane S. Lucas; Claudia E. Kuehni |
| | DOI | doi:10.1183/13993003.01181-2016 |
| | Type | Journal article |
| | Published in | European Respiratory Journal |
| | Published by | European Respiratory Society (ERS) |
| | ISSNs | 0903-1936 ; 1399-3003 |
| | Subject | Pulmonary and Respiratory Medicine |
| | Link | https://syndication.highwire.org/content/doi/10.1183/13993003.01181-2016 |
| 24. | doi:10.1183/13993003.01090-2016 Title | European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia |
| | Authors | Jane S. Lucas; Angelo Barbato; Samuel A. Collins ; Myrofora Goutaki; Laura Behan; Daan Caudri; Sharon Dell; Ernst Eber; Estelle Escudier; Robert A. Hirst; Claire Hogg; Mark Jorissen; Philipp Latzin; Marie Legendre; Margaret W. Leigh; Fabio Midulla; Kim G. Nielsen; Heymut Omran; Jean-Francois Papon; Petr Pohunek; Beatrice Redfern; David Rigau; Bernhard Rindlisbacher; Francesca Santamaria; Amelia Shoemark; Deborah Snijders; Thomy Tonia; Andrea Titieni; Woolf T. Walker; Claudius |

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| | | Werner; Andrew Bush; Claudia E. Kuehni |
| | DOI | doi:10.1183/13993003.01090-2016 |
| | Type | Journal article |
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| | Subject | Pulmonary and Respiratory Medicine |
| | Link | https://syndication.highwire.org/content/doi/10.1183/13993003.01090-2016 |
| 25. | doi:10.1183/13993003.00736-2016 | Title |
| | | Clinical manifestations in primary ciliary dyskinesia: systematic review and meta-analysis |
| | Authors | Myrofora Goutaki; Anna Bettina Meier; Florian S. Halbeisen; Jane S. Lucas; Sharon D. Dell; Elisabeth Maurer; Carmen Casaulta; Maja Jurca; Ben D. Spycher; Claudia E. Kuehni |
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| | ISSNs | 0903-1936 ; 1399-3003 |
| | Subject | Pulmonary and Respiratory Medicine |
| | Link | https://syndication.highwire.org/content/doi/10.1183/13993003.00736-2016 |
| 26. | doi:10.1183/13993003.01551-2015 | Title |
| | | PICADAR: a diagnostic predictive tool for primary ciliary dyskinesia |
| | Authors | Laura Behan; Borislav D. Dimitrov; Claudia E. Kuehni; Claire Hogg; Mary Carroll; Hazel J. Evans; Myrofora Goutaki; Amanda Harris; Samantha Packham; Woolf T. Walker; Jane S. Lucas |
| | DOI | doi:10.1183/13993003.01551-2015 |
| | Type | Journal article |
| | Published in | European Respiratory Journal |
| | Published by | European Respiratory Society (ERS) |
| | ISSNs | 0903-1936 ; 1399-3003 |
| | Subject | Pulmonary and Respiratory Medicine |
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- [content/doi/10.1183/13993003.01551-2015](https://doi.org/10.1183/13993003.01551-2015)
27. [doi:10.1590/1806-3713/e20170358](https://doi.org/10.1590/1806-3713/e20170358) Title
- Translation of the quality-of-life measure for adults with primary ciliary dyskinesia and its application in patients in Brazil
- Authors [Ana Paula Lima de Queiroz](#); [Rodrigo Abensur Athanazio](#); [Mary Anne Kowal Olm](#); [Bruna Rubbo](#); [Yuri Reis Casal](#); [Jane Lucas](#); [Laura Behan](#)
- DOI [doi:10.1590/1806-3713/e20170358](https://doi.org/10.1590/1806-3713/e20170358)
- Type Journal article
- Published in Jornal Brasileiro de Pneumologia
- Published by FapUNIFESP (SciELO)
- ISSN [1806-3756](#); [1806-3713](#)
- Subject Pulmonary and Respiratory Medicine
- Link <http://www.scielo.br/pdf/jbpneu/v45n3/1806-3713-jbpneu-45-03-e20170358.pdf>
28. [doi:10.1007/s11136-017-1564-y](https://doi.org/10.1007/s11136-017-1564-y) Title
- The patient's experience of primary ciliary dyskinesia: a systematic review
- Authors [Laura Behan](#); [Bruna Rubbo](#); [Jane S. Lucas](#); [Audrey Dunn Galvin](#)
- DOI [doi:10.1007/s11136-017-1564-y](https://doi.org/10.1007/s11136-017-1564-y)
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- Published in Quality of Life Research
- Published by Springer Nature
- ISSN [0962-9343](#); [1573-2649](#)
- Links <http://link.springer.com/article/10.1007/s11136-017-1564-y/fulltext.html>; <http://link.springer.com/content/pdf/10.1007/s11136-017-1564-y.pdf>
29. [doi:10.1136/thoraxjnl-2016-209356](https://doi.org/10.1136/thoraxjnl-2016-209356) Title
- Validation of a health-related quality of life instrument for primary ciliary dyskinesia (QOL-PCD)
- Authors [Laura Behan](#); [Margaret W Leigh](#); [Sharon D Dell](#); [Audrey Dunn Galvin](#); [Alexandra L Quittner](#); [Jane S Lucas](#)
- DOI [doi:10.1136/thoraxjnl-2016-209356](https://doi.org/10.1136/thoraxjnl-2016-209356)
- Type Journal article
- Published in Thorax
- Published by BMJ
- ISSN [0040-6376](#); [1468-3296](#)
- Link <https://syndication.highwire.org/>

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30. [doi:10.1183/13993003.02018-2015](https://doi.org/10.1183/13993003.02018-2015)Title
- Diagnosing primary ciliary dyskinesia: an international patient perspective
- Authors Laura Behan; Audrey Dunn Galvin; Bruna Rubbo; Sarah Masfield; Fiona Copeland; Michele Manion; Bernhard Rindlisbacher; Beatrice Redfern; Jane S. Lucas
- DOI [doi:10.1183/13993003.02018-2015](https://doi.org/10.1183/13993003.02018-2015)
- Type Journal article
- Published in European Respiratory Journal
- Published by European Respiratory Society (ERS)
- ISSNs [0903-1936](https://doi.org/10.1183/13993003.02018-2015); [1399-3003](https://doi.org/10.1183/13993003.02018-2015)
- Subject Pulmonary and Respiratory Medicine
- Link <https://syndication.highwire.org/content/doi/10.1183/13993003.02018-2015>
31. [doi:10.1183/13993003.00749-2015](https://doi.org/10.1183/13993003.00749-2015)Title
- Accuracy of diagnostic testing in primary ciliary dyskinesia
- Authors Claire L. Jackson; Laura Behan; [Samuel A. Collins](https://doi.org/10.1183/13993003.00749-2015); Patricia M. Goggin; Elizabeth C. Adam; Janice L. Coles; Hazel J. Evans; Amanda Harris; Peter Lackie; Samantha Packham; Anton Page; James Thompson; Woolf T. Walker; Claudia Kuehni; Jane S. Lucas
- DOI [doi:10.1183/13993003.00749-2015](https://doi.org/10.1183/13993003.00749-2015)
- Type Journal article
- Published in European Respiratory Journal
- Published by European Respiratory Society (ERS)
- ISSNs [0903-1936](https://doi.org/10.1183/13993003.00749-2015); [1399-3003](https://doi.org/10.1183/13993003.00749-2015)
- Subject Pulmonary and Respiratory Medicine
- Link <https://syndication.highwire.org/content/doi/10.1183/13993003.00749-2015>
32. [doi:10.1183/09031936.00216214](https://doi.org/10.1183/09031936.00216214)Title
- A quality-of-life measure for adults with primary ciliary dyskinesia: QOL-PCD
- Authors Jane S. Lucas; Laura Behan; Audrey Dunn Galvin; Adrienne Alpern; Anjana M. Morris; Mary P. Carroll; Michael R. Knowles; Margaret W. Leigh; Alexandra L. Quittner

DOI [doi:10.1183/09031936.00216214](https://doi.org/10.1183/09031936.00216214)
 Type Journal article
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 ISSN [0903-1936](https://doi.org/10.1183/09031936.00216214); [1399-3003](https://doi.org/10.1183/09031936.00216214)
 Subject Pulmonary and Respiratory Medicine
 Link <https://syndication.highwire.org/content/doi/10.1183/09031936.00216214>

33. [doi:10.1038/ncomms14279](https://doi.org/10.1038/ncomms14279)Title

Authors

X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3
 Chiara Olcese; Mitali P. Patel; Amelia Shoemark; Santeri Kiviluoto; Marie Legendre; Hywel J. Williams; Cara K. Vaughan; Jane Hayward; Alice Goldenberg; Richard D. Emes; Mustafa M. Munye; Laura Dyer; Thomas Cahill; Jeremy Bevallard; Corinne Gehrig; Michel Guipponi; Sandra Chantot; Philippe Duquesnoy; Lucie Thomas; Ludovic Jeanson; Bruno Copin; Aline Tamalet; Christel Thauvin-Robinet; Jean- François Papon; Antoine Garin; Isabelle Pin; Gabriella Vera; Paul Aurora; Mahmoud R. Fassad; Lucy Jenkins; Christopher Boustred; Thomas Cullup; Mellisa Dixon; Alexandros Onoufriadis; Andrew Bush; Eddie M. K. Chung; Stylianos E. Antonarakis; Michael R. Loebinger; Robert Wilson; Miguel Armengot; Estelle Escudier; Claire Hogg; Saeed Al-Turki; Carl Anderson; Dinu Antony; Inês Barroso; Philip L. Beales; Jamie Bentham; Shoumo Bhattacharya; Keren Carss; Krishna Chatterjee; Sebahattin Cirak; Catherine Cosgrove; Daly Allan; Richard Durbin; David Fitzpatrick; Jamie Floyd; A. Reghan Foley; Chris Franklin; Marta Futema; Steve E. Humphries; Matt Hurles; Shane McCarthy; Dawn Muddyman; Francesco Muntoni; Victoria Parker; Felicity Payne; Vincent Plagnol; Lucy Raymond; David B. Savage; Peter J. Scambler; Miriam Schmidts; Robert Semple; Eva Serra; Jim Stalker; Margriet van

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| <p>ISSN Links</p> | <p>1471-2466 http://link.springer.com/content/pdf/10.1186/s12890-016-0261-x.pdf; http://link.springer.com/article/10.1186/s12890-016-0261-x/fulltext.html; http://link.springer.com/content/pdf/10.1186/s12890-016-0261-x</p> |
| <p>36. doi:10.1080/01913123.2017.1362089Title</p> | <p>Should transmission electron microscopy and ultrastructural cilia evaluation remain part of the diagnostic work-up for primary ciliary dyskinesia? Claudius Werner; Panayiotis Kouis DOI doi:10.1080/01913123.2017.1362089 Type Journal article Published in Ultrastructural Pathology Published by Informa UK Limited ISSN 0191-3123; 1521-0758 Link https://www.tandfonline.com/doi/pdf/10.1080/01913123.2017.1362089</p> |
| <p>37. doi:10.1183/13993003.00776-2015Title</p> | <p>An international registry for primary ciliary dyskinesia Claudius Werner; Martin Lablans; Maximilian Ataian; Johanna Raidt; Julia Wallmeier; Jörg Große-Onnebrink; Claudia E. Kuehni; Eric G. Haarman; Margaret W. Leigh; Alexandra L. Quittner; Jane S. Lucas; Claire Hogg; Michal Witt; Kostas N. Priftis; Panayiotis Yiallourous; Kim G. Nielsen; Francesca Santamaria; Frank Ückert; Heymut Omran DOI doi:10.1183/13993003.00776-2015 Type Journal article Published in European Respiratory Journal Published by European Respiratory Society (ERS) ISSN 0903-1936; 1399-3003 Subject Pulmonary and Respiratory Medicine Link https://syndication.highwire.org/content/doi/10.1183/13993003.00776-2015</p> |
| <p>38. doi:10.1016/j.ajhg.2015.08.012Title</p> | <p>Loss-of-Function GAS8 Mutations Cause Primary Ciliary Dyskinesia and Disrupt the Nexin-Dynein Regulatory Complex</p> |

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|--------------|---|
| Authors | Heike Olbrich; Carolin Cremers; Niki T. Loges; Claudius Werner; Kim G. Nielsen; June K. Marthin; Maria Philipsen; Julia Wallmeier; Petra Pennekamp; Tabea Menchen; Christine Edelbusch; Gerard W. Dougherty; Oliver Schwartz; Holger Thiele; Janine Altmüller; Frank Rommelmann; Heymut Ocran |
| DOI | doi:10.1016/j.ajhg.2015.08.012 |
| Type | Journal article |
| Published in | The American Journal of Human Genetics |
| Published by | Elsevier BV |
| ISSN | 0002-9297 |
| Links | https://api.elsevier.com/content/article/PII:S0002929715003341?httpAccept=text/plain; https://api.elsevier.com/content/article/PII:S0002929715003341?httpAccept=text/xml |

39. [doi:10.1183/13993003.00314-2017](https://doi.org/10.1183/13993003.00314-2017)Title

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|--------------|---|
| Authors | Infertility in an adult cohort with primary ciliary dyskinesia: phenotype–gene association Gert Jan Vanaken; Laurence Bassinet; Mieke Boon; Rahma Mani; Isabelle Honoré; Jean-Francois Papon; Harry Cuppens; Martine Jaspers; Natalie Lorent; André Coste; Estelle Escudier; Serge Amselem; Bernard Maitre; Marie Legendre; Sophie Christin-Maitre |
| DOI | doi:10.1183/13993003.00314-2017 |
| Type | Journal article |
| Published in | European Respiratory Journal |
| Published by | European Respiratory Society (ERS) |
| ISSNs | 0903-1936 ; 1399-3003 |
| Subject | Pulmonary and Respiratory Medicine |
| Link | https://syndication.highwire.org/content/doi/10.1183/13993003.00314-2017 |

Projects

The Action reported 0 project(s) and 2 proposal(s) resulting from the Action networking.

"N/A"

Other outputs / achievements

The following other outputs/ achievements contributing to the COST mission resulted from the Action:

1. Partners have been successful obtaining local/ national funding to support the Actions' aims. For example

1. University of Cyprus internal funding (50000 €) for the implementation of research project: "Development of air liquid interface cell cultures of ciliated respiratory epithelium for assessment of mucociliary clearance defects in vitro"
 2. University of Southampton, UK and the UK diagnostic centres: NIHR RfPB: PB-PG-1215-20014 – Accuracy of high-speed video microscopy analysis to diagnose primary ciliary dyskinesia
 3. University of Southampton: Funding from NIHR BRC to investigate *Haemophilus influenzae* biofilm infection in PCD (2 PhD students)
 4. The networking between Institute of Human Genetics PAS in Poland and the University Hospital of Southampton, UK, led to a successful application for a fellowship for a PhD student from Poland, Maciej Dabrowski, MSc (grant of the Polish Ministry of Science) to finance a five-month stay at the laboratory of Prof. Jane Lucas in Southampton, UK. This allows an in-depth training of the Polish ECR in the diagnostic procedures and scientific techniques used in the process of PCD diagnosis in the PCD National Service, UK. After the return to Poland, this knowledge will be introduced into the home laboratory.
 5. University of Bern, Institute of Social and Preventive Medicine, Switzerland: Swiss National Science Foundation 320030_173044_1: Natural history, phenotypes and disease classification in primary ciliary dyskinesia
-
2. Portuguese patients and their carers met with members of the UK PCD Support Group and with clinicians during the BEATPCD conference in Lisbon. This was the first meeting in a move to set up a Patient Support group in Portugal.

Impacts

The Action reported the following impact(s):

| Description of the impact, i.e. what will change, and for whom, as a result of what the Action achieved | Type of impact | Timing of impact |
|--|--|---------------------------|
| Accelerated understanding of PCD genetics. A number of new genes have been discovered and characterised through collaborations of BEAT-PCD clinicians and researchers. These discoveries are already impacting PCD diagnostic testing. Moving forward our improved knowledge of the underlying genetics and pathophysiological mechanisms of PCD will provide potential target therapies for clinical trials. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| Sustainability of clinical care and research development for PCD. We have grown the capacity of early career researchers and clinicians, both in the quantity of people and the quality of their knowledge, skills and networking capabilities. There is excitement amongst the early and mid- career group about continuing to investigate and manage this rare disease. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| A consensus statement for TEM as a diagnostic tool has provided standardised terminology and reporting. This was achieved through workshops and e-surveys. We validated the recommendations within the BEAT-PCD network and these are now being followed in many PCD centres across Europe. The impact is improved standardisation of a clinical diagnosis and of reporting. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| A consensus statement for defining pulmonary exacerbations has been published. The impact is an outcome for use in clinical trials. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| Several countries have significant changes to diagnostic standards and accessibility as a direct consequence of training and networking through the BEATPCD COST Action network. For example, the MC member from Norway reports ".....as a result of this Action, PCD patients in Norway have access to an improved diagnostic service. The Action has enabled the training of at least 1 adult physician in PCD diagnosis and management (not enough I know) and the establishment of an adult PCD clinic which will serve the whole country for diagnostics and annual follow-up. It has facilitated me getting a small grant of 50,000 euro to establish a Norwegian PCD register and employ a short-term research fellow to conduct basic epidemiological research on Norwegian patients. This will lay the groundwork for an application for a grant to support a PhD student. None of this would have been possible without the wonderful BEAT-PCD. I really hope it can continue in some form". | <ul style="list-style-type: none"> Scientific / Technological Societal | Achieved |
| Sharing of in vivo and ex vivo models for pre-clinical trials of novel therapies. This has mostly occurred through STSMs. The impact is better research in more laboratories. The impact is already seen, and will increase as the sharing extends. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| Evidence based guidelines for the diagnosis of PCD have been published and are now in use in most European Reference centres (European Respiratory Society). The impact is a better and more standardised approach to diagnosing PCD. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| A European network of PCD, clinicians delivering expert care through ERN-Lung reference centres across Europe https://ern-lung.eu/ The PCD core group formed directly from meetings and discussions of the BEATPCD Cost Action. | <ul style="list-style-type: none"> Scientific / Technological | Achieved |
| Standardised proforma for the collection of clinical data across Europe. The proforma is in a late stage of piloting and will soon be ready for general use in clinics. The impact will be a tool that can be | <ul style="list-style-type: none"> Scientific / Technological | Foreseen within two years |

| | | |
|--|--|----------------------------------|
| <p>used for international clinical trials and research. It will also improve data collection at a local level.</p> | | |
| <p>A review of outcome measures for use in clinical trials in PCD has been completed. Two reports are now being collated for publication (1. spirometry 2. outcomes other than spirometry). We will then plan a statement on how to use outcome measures in clinical trials.</p> | <ul style="list-style-type: none"> • Scientific / Technological | <p>Foreseen within two years</p> |
| <p>A consensus statement for preventing cross infection is at a late stage following a series of BEAT-PCD workshops and e-surveys. The impact will be standardised international guidance for healthcare professionals and patients, leading to improved health outcomes.</p> | <ul style="list-style-type: none"> • Scientific / Technological | <p>Foreseen within two years</p> |
| <p>A Europe wide research network positioned to obtain platform grants for treatment of PCD. The connections between clinicians and scientists has been particularly impactful in BEATPCD. We are now understanding each other's 'language' and a number of small projects are in place, with plans for large projects building on the preliminary data generated.</p> | <ul style="list-style-type: none"> • Scientific / Technological | <p>Foreseen within two years</p> |

Dissemination and exploitation of Action results

Dissemination and exploitation approach of the Action

The Action's dissemination and exploitation approach as well as all activities undertaken to ensure dissemination and exploitation of Action results and the outcomes of these activities are described below.

We have used a project management tool, Basecamp to ensure with have good communication and dissemination with the membership. We have a website which is targeted at all stake holders and the general public <https://www.beatpcd.org/> We ensure that BEAT publications are cited on our website <https://www.beatpcd.org/publications> We contribute to Open Access fees for key publications from the network. A number of members are actively involved in their patient organisations and keep them updated about BEATPCD. Patient representatives have participated in all Action conferences, and training schools. They have been participants in most projects arising from the Action. We have used social media, in particular twitter @beatpcd Our account is particularl active during conferences; we have a growing number of followers from outside the network

Dissemination meetings funded by the Action

The Action did not fund any Dissemination Meetings

Other dissemination activities

The Action also undertook the following dissemination activities:

| | |
|-----------------|--|
| Activity | EMBO Cilia 2018 Conference: Copenhagen, Denmark, Jane Lucas delivered a symposium lecture: Diagnosing patients with primary ciliary dyskinesia |
| Target | An international audience of scientists primarily focusing their research on primary cilia, and some focusing on motile cilia. |
| Outcome | Dissemination about clinical aspects of primary ciliary dyskinesia to primary cilia experts. |
| Link | http://meetings.embo.org/event/18-cilia |

| | |
|-----------------|--|
| Activity | European Respiratory Society Congress 2018. Jane Lucas delivered a symposium lecture: Paediatric State of the Art Session: Primary Ciliary Dyskinesia |
| Target | Trainee, Policy maker, Clinical researcher, Critical/Intensive care physician, Nurse, Scientist (basic, translational), Paediatrician, Patient, Physiologist, Physiotherapist, Student, General practitioner, Lung function technologist |
| Outcome | Dissemination of information about diagnosing and treating PCD to a wide audience |
| Link | https://erscongress.org/programme-2018/access-the-programme-2018.html |

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|-----------------|--|
| Activity | European Respiratory Society Congress 2018 Symposium "Recent advances in primary ciliary dyskinesia" The chairs and speakers were members the Action BEATPCD: Chairs B Karadag (Turkey), J Lucas (UK); Speakers: H Omran (Germany) E Haarman (Netherlands), C Kuehni (Switzerland) |
| Target | Trainee, Policy maker, Adult pulmonologist/Clinician, Clinical researcher, Nurse, Paediatrician, Patient, Physiotherapist, Student, General practitioner, Lung function technologist |
| Outcome | Dissemination to a large symposium of recent advances in PCD, many arising from the activities of BEATPCD. |
| Link | https://erscongress.org/programme-2018/access-the-programme-2018.html |

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|-----------------|--|
| Activity | ERS Congress 2018: A Thematic Poster session "Thematic poster Insights in primary ciliary dyskinesia, asthma and lung function testing" and a Poster discussion session "Primary ciliary dyskinesia and lung |
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| | function tests in respiratory morbidities" |
| Target | Epidemiology, General respiratory patient care, Physiology, Public health, Cell and molecular biology. |
| Outcome | Both poster sessions were dominated by research from the Action, BEATPCD. The research was mostly presented by early career researchers, and two ECRs were co-chairs. |
| Link | https://erscongress.org/programme-2018/access-the-programme-2018.html |

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|-----------------|---|
| Activity | 7th International Meeting on Pulmonary Rare Diseases and Orphan Drugs- Jane Lucas spoke about Primary ciliary dyskinesia. 2018 |
| Target | Predominantly adult and paediatric pulmonologists working in the field of rare lung disease. |
| Outcome | Networking and dissemination opportunity with clinicians working in other rare diseases. |
| Link | http://www.ilpolmone.it/international-meeting-on-pulmonary-rare-diseases-and-orphan-drugs |

| | |
|-----------------|---|
| Activity | Kongress der Deutschen Gesellschaft für Pneumologie und Beatmungsmedizin e.V. (59th German Pulmomonology Congress) Symposium «Ciliary Dysfunktion» H. Omran (Germany) and M.Goutaki (Switzerland) spoke about Diagnosis and Treatment of PCD and about PCD epidemiology. 2018 |
| Target | The target audience included: Trainee, Adult pulmonologist, Clinicians, Clinical researchers, |
| Outcome | The event provided a Networking and dissemination opportunity with adult pulmonologists |
| Link | https://pneumologie-kongress.de/ |

| | |
|-----------------|---|
| Activity | Kartagener Syndrom & Primäre Ciliäre Dyskinesie e.V. Patientenkongress (Congress of the German speaking PCD patient support group) Several members of BEAT-PCD spoke about research on primary ciliary dyskinesia and management (including H.Omran, M.Goutaki, A.Jung, C. Koerner-Rettberg) 2018 |
| Target | The event provided an opportunity for clinical members of BEATPCD to explain activities of our network to patients. |
| Outcome | The activities of BEATPCD COST Action are better understood by patients. Patients have an opportunity to get involved in the COST Action, to participate and advise about research and clinical priorities. |
| Link | https://www.kartagener-syndrom.org/ |

| | |
|-----------------|--|
| Activity | Jahresversammlung der Schweizerischen Gesellschaft für Pneumologie (SGP) und Schweizerischen Gesellschaft für Pädiatrische Pneumologie (SGPP) (Annual congress of Swiss pulmonology and paediatric pulmonology) Symposium on PCD. Chairs (A.Jung) and speakers (C.Kuehni, P. Latzin, S. Blanchon) were members of the action |
| Target | The target audience included: Trainees, Adult pulmonologists and Clinicians, Clinical researchers, Paediatricians |
| Outcome | Networking and dissemination opportunity with adult pulmonologists and paediatricians, providing an opportunity to increase knowledge about PCD amongst non-specialists. |
| Link | https://www.pneumo-congress.ch/sgp2019.html |

| | |
|-----------------|---|
| Activity | ERN-LUNG Kick-Off Meeting in Frankfurt, April 2017 – Heymut Omran presented activities of the PCD-Core including BEATPCD Activities |
| Target | Adult and paediatric pulmonologists and researchers working in the field of rare lung disease. |
| Outcome | Networking and dissemination opportunity with clinicians and researchers working in other rare diseases |
| Link | https://ern-lung.eu/ |

Exploitation activities

The Action undertook the following activities to ensure exploitation (use, in particular in a commercial context) of the Action's achievements:

No exploitation activities were reported by the Action.

Action Success(es)

The Action's two most significant successes were the following:

- A direct outcome of this network has led to the accreditation of 10 PCD centres as members of the European Reference Network on rare lung diseases (ERN-LUNG). ERN-LUNG has been established to provide an opportunity for clinicians to work together in Europe in order to tackle the challenge of rare diseases. ERN-LUNG will support clinicians and researchers to share expertise, knowledge and resources across the EU, and ensure doctors have the most recent and expert knowledge possible to ensure excellent treatment and care. ERN-LUNG is a project of building a clinical care network for all rare diseases of the respiratory system. ERN-LUNG has been evaluated by the European Commission and on 15 December 2016 the European Reference Network Board of Member States approved ERN-LUNG as one of the 24 ERNs for rare disease. PCD is a core group within ERN-LUNG, spinning out from the Action. ERN-LUNG PCD group was initially discussed and planned during BEATPCD Cost Action MC meetings, with an invited speaker from Eurordis; since successful commissioning of ERN-LUNG, the PCD core group has met in a public and inclusive session during BEATPCD conferences in 2017, 18 and 19 and expanded with currently 1 affiliated and 4 supporting partners.
- ERS Diagnostic Guidelines for the diagnosis of PCD: members of BEAT-PCD contributed to development and dissemination of this important evidence based guideline which will ensure patients and participants in trials have a standardised diagnosis. The guideline has been published in European Respiratory Journal, and presented at the ERS congress in a symposium session. Further dissemination resources are being prepared including a CME online training module, a slide-set, and a pocket-book summary. At the BEATPCD meeting in Paris 2018 we invited authors of the North American guideline with an aim to resolving differences which are mostly minor; this has resulted in an invited manuscript co-authored by European and North Americans which will be published in 2019. Having identified areas for standardisation of diagnostic methods during the development of the guideline, Action BEATPCD has developed and validated guidelines for analysing and reporting TEM.

Action Expenditure

The table below shows the budget allocated to the Action for each Grant Period:

| # | Grant Period | Start Date | End Date | Budget allocated to Action (EUR) |
|---|--------------|------------|-----------|----------------------------------|
| 1 | CGA-BM1407-1 | 1-7-2015 | 30-4-2016 | 127,040.50 (EUR) |
| 2 | AGA-BM1407-2 | 1-5-2016 | 30-4-2017 | 105,938.00 (EUR) |
| 3 | AGA-BM1407-3 | 1-5-2017 | 30-4-2018 | 124,995.46 (EUR) |
| 4 | AGA-BM1407-4 | 1-5-2018 | 30-4-2019 | 130,674.50 (EUR) |