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| I. Project Title and pCode | |
| Genes related to Genetic And Rare lung Diseases (GARD)  as risk for LC | **Date: 2023-04-11 (2018-12-03)** |
| **II. Project Leaders (List Names and Institutes)** | **Primary Contact**  **Information** |
| Albert Rosenberger, Ph.D., UMG  Heike Bickeböller, Ph.D. , UMG  Christopher Amos, Ph.D. , Baylor College | **Name:**  Dr. Albert Rosenberger  **Email:**  [**arosenb@gwdg.de**](mailto:arosenb@gwdg.de)  **Tel:**  **++49 (551) 39-1404** |
| III. Progress to date. *What has been done so far to move the project forward (data pooling, grant submission, etc), and when it was done* | |
| The main goal of the project is to investigate possible associations of all genes related to GARDs with LC. We originally based our gene selection on the collection of the US-NIH “Genetic and Rare Diseases (GARD) Information Center” (<https://rarediseases.info.nih.gov/>). This collection was updated and restructures in the last two years. We also recognized the exchange of records with and among other relevant public data sources:ORPHANET, MONARCH, OMIM. In addition, query options or updated data sets from these databases are now available. As a result, we had to redesign the project basis.  We were also expanding the definition of "gene" by adding enhancer regions, which are now robustly available and have been shown to increase performance for gene-based analysis. | |
| **IV. Project Status and Preliminary Results**. *S****ummarize the current status of the project. Report preliminary results if there is any*** | |
| Due to this further development of the basic information sources, we had to "restart" the project in early 2022.  There are no preliminary results.  Currently, we implement our routines on the cluster system of the Baylor College.  NOTE: The proposal replaces two (former) proposals:   1. Lung cancer risk among carriers of SERPINA1 variants,  by Julia Heck (3 Sept 2016, later withdrawn) 2. SERPINA1-induced Alpha-1 antitrypsin deficiency as a risk for lung cancer by Albert Rosenberger (12 July 2018)   Dr. Julia Heck did not at all participated in the implementation of the project. Hence, she is no more listed as project leader. | |
| **V. Justification of Extension** | |
| Delay due to progress in the basic information sources. | |
| **VI. Future Plans & Timeline** | |
| I guess, we will need (at least) another year to go on this project to achiev reliable resuslts. | |