Current status of the Asthma and Allergy Database

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ABSTRACT:

The database provides an online resource for access to data on the genetics of asthma and allergy. This report describes the present status of the site. Currently, a detailed description of 88 linkage studies (7164 linkage positions) and 72 mutation studies are available. The results can be accessed in table form or graphically. The database also contains mouse asthma studies and human homology relationships, gene expression studies and links to relevant patents. Technical details about the server architecture, database installation, database construction, database structure and the user interface are explained elsewhere [Wjst and Immervoll (1998) *Bioinformatics*, 14, 827–828]. The URL is http://cooke.gsf.de

INTRODUCTION

Asthma is one of the most frequent chronic diseases affecting now 5–10% of the population in Western countries. Up to 30% of children are reporting asthma symptoms (1). It is clinically characterized by chronic, intermittent airway obstruction with wheeze, cough and breathnessless. A familial clustering of asthma has been known for a long time (2), however, the inheritance does not follow a simple Mendelian pattern. Starting as early as 1985 we are currently experiencing an explosive increase of mutation and linkage studies for allergic diseases and asthma. To get an overview on this wealth of information presented at several meetings and in many journals is getting more and more difficult. Therefore an internet database seems to be a new possibility to cope with this huge amount of information.

DESCRIPTION OF THE DATABASE:

The user can reach the asthma and allergen database at the URL http://cooke.gsf.de . The main database is divided into linkages and mutations. As of July 27, 1998, the database covered 88 human linkage studies (7164 linkage positions) and 72 mutation studies recorded from 88 different papers (Table 1). The user can look for each chromosome or the whole genome in a table or a graphical figure. Moreover it is possible to search for references, phenotypes or populations entered in the database. The linkage studies give information about chromosomal locations, linked

 Table 1. Summary of data provided by the asthma and allergy gene database

| | Number |
|---|--------|
| Linkage studies | 88 |
| Mutation studies (24 genes) | 72 |
| Gene expression studies | 1 |
| Mouse linkage studies | 3 |
| Published papers | 88 |
| Populations from different regions | 18 |
| Phenotypes | 54 |
| Included families (partially redundant as | 5 005 |
| included in several studies) | |
| Included subjects (partially redundant as | 32 605 |
| included in several studies) | |

markers, P-values, phenotypes, number of probands/families, populations, used statistical methods and references. The mutation study section gives advice on the mutated gene, its chromosomal location, the cytogenetic position, the type of mutation (mutated and normal DNA or RNA or protein sequence), the mutation symbol, whether an exon, an intron or the promoter of a gene is mutated, the detection method of the mutation, the inheritance mode and additional study information comparable to the linkage studies (Fig. 1). There are also minor sections, which contain at the time of submission three different mouse genome screens of asthma related traits with human mouse homology relationships. As the identification of relevant genes should not only be possible by classical linkage or association studies but also by observing differing expression patterns, studies of gene expression in asthmatic patients are also collected. It is also possible to look for more asthma references, such as reviews (32 entries), books (6 entries), supplements (3 entries) as well as information on segregation (14 entries), twin (17 entries) and family studies (41 entries), all dealing with asthma genetics. In the patents section there are links to ~500 DNA patents related to asthma and allergy, which may be useful for scientists studying asthma candidate genes. All submitted data are checked by both authors to optimize data queries.

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| Lesezeichen | & Netste: http://cooke.gsf.de/asthmaGen/ | |
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| SF National esearch Center | Gene Symbol: beta2AR | |
| esearch Center r Environment | Gene Name: beta2-adrenoceptor | |
| nd Health | Gene Name: beta2AR | |
| stitute of | Location: q31 | |
| oidemiology | Chromosome: 05 | |
| | Exon/Intron: exon | |
| troduction | Exon/Intron Number: 1 | |
| | Mutation Symbol: missing | |
| gistration | Mutation Synonym: Arg16Gly | |
| embers | Long Description: A to G substitution resulting in a Arg to Gly exchange in the beta2AR protein | n |
| ailing list | EMBL ID: not known | |
| onference | Starts at 0, ends at 0 (lenght 0) | |
| | DNA normal sequence: 46A | |
| ene links | DNA mutated sequence: 46G | |
| thma links | mRNA normal sequence: 46A | |
| ouse page | mRNA mutated sequence: 46G | |
| | Protein normal sequence: 16Arg | |
| ne expression | Protein mutated sequence: 16Gly | |
| ferences | Detection Method: Ncol digest | |
| atents | Confirmation Method: sequencing 5'-Primer: 5'-GCCTTCTTGCTGGCACCCCAT-3' | |
| nkages & | 3'-Primer: 5'-CAGACGCTCGAACTTGGCCATG-3' | |
| utations | New Restriction Site: Ncol | |
| litor | Lost Restriction Site: not known | |
| MEUT | Variant type: polymorphism | |
| Munich, Germany | Event: substitution | |
| 18 A | Novelty: not known | |
| | Inheritance Mode: autosomal | |
| | % of population positive: missing | |
| | Relative Risk: missing | |
| | Promoting Factor: missing | 1 |

Figure 1. Detailed description of a selected mutation in the candidate gene β -2 adrenoceptor on chromosome 5q31.

Technical details

Technical details about the server architecture, website installation, database construction, database structure and the user interface are explained elsewhere (3).

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