

# 腫瘍領域の 遺伝カウンセリング



# 腫瘍領域のGC @イギリス (シェフィールド)

## ■ 状況

- ハイリスクの人のみがGenetic Centreに紹介される
- 遺伝学的検査を受検するには遺伝カウンセリングは必須
- 乳がんの相談が最も多い

## ■ 遺伝学的検査

- single gene sequencing, パネルは稀
- 陰性だった場合に、他の検査(MLPA)、研究への参加等を検討
- in house labで検査を行うことが多い
- variantの評価はin house labのバイオインフォマティクスが行う
- variantの情報はDECIPHERから得る。新規の場合はDECIPHERに変異情報と臨床情報を登録する。

## ■ これまでの実績

- 発症者に対する遺伝カウンセリング 件数：約500件/年間
- 血縁者に対する遺伝カウンセリング 件数(発端者変異同定済)：約120-150件/年間



# 腫瘍領域のGC @イギリス (シェフィールド)

## ■ 遺伝カウンセリングの流れ

- (1) 紹介状
- (2) ミーティングで方針決定
- (3) 家族歴の質問票送付, 家系図作成
- (4) (必要に応じてミーティング)
- (5) 初診・採血
- (6) 結果開示, 状況に合わせて対面/電話を選択
- (7) (病的変異が同定された人に対してフォローアップ)

## ■ 各スタッフの役割

- 医師(clinical geneticist) : 発端者の遺伝学的診断, 医学的な管理
- 遺伝カウンセラー(RGC) : 家系図の確認→遺伝的リスクの評価  
遺伝カウンセリング時に必要な資料の作成  
血縁者の遺伝カウンセリング, 結果開示 :  
陽性者のフォローアップ電話
- Administrator : 外来予約, 返書のタイピング
- Coordinator : 家族歴質問票の送付, 家系図の作成



# 乳がんのGC @イギリス (シェフィールド)

## ■ 乳がん

- NICE guidelines : 診断, サーベイランスなどすべてこのガイドラインに従っている
- リスク算定 : Manchester Scoreを使用。BOADICEAを使用することもある  
Manchester Score で15%以上であると、変異が見つかるのは10%  
50y以下であればスコアの値によらずGTを行う
- GTの方針 : single gene sequencing, MLPAは同時には行わない  
パネルを使用しないのはBRCA以外はmoderate risk geneであり  
臨床的有用性が不十分と判断しているため  
30-40yは乳腺がdenseなのでBRCA以外にTP53もみてマンモグラフィー  
が可能か判断
- 病的変異が認められた場合: breast care teamでフォロー, 卵巣は婦人科でフォロー  
前立腺はGPがフォロー  
すべてのサーベイランス, RRSO 無償 (国が負担)

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# Centre for Cancer Genetic Epidemiology

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## BOADICEA

### BOADICEA

- > [BOADICEA model description](#)
- > [BOADICEA Web Application](#)
- > [Setup your BOADICEA user account](#)
- > [Login to BWA v3](#)
- > [Login to BWA v4 Beta](#)
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The **Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm** (BOADICEA) is a computer program that is used to calculate the risks of breast and ovarian cancer in women based on their family history. It is also used to calculate the probability that they are carriers of cancer-associated mutations in the BRCA1 or BRCA2 gene. To access BOADICEA, all you need is a BOADICEA user account, which you can setup online in a minutes [here](#).

You can access two different versions of the BOADICEA program using the links in the menu to the left:

- (i) BWA v3 considers the explicit effects of BRCA1 and BRCA2 mutations;
- (ii) BWA v4 *Beta* considers the explicit effects of BRCA1, BRCA2, PALB2, CHEK2 and ATM mutations.

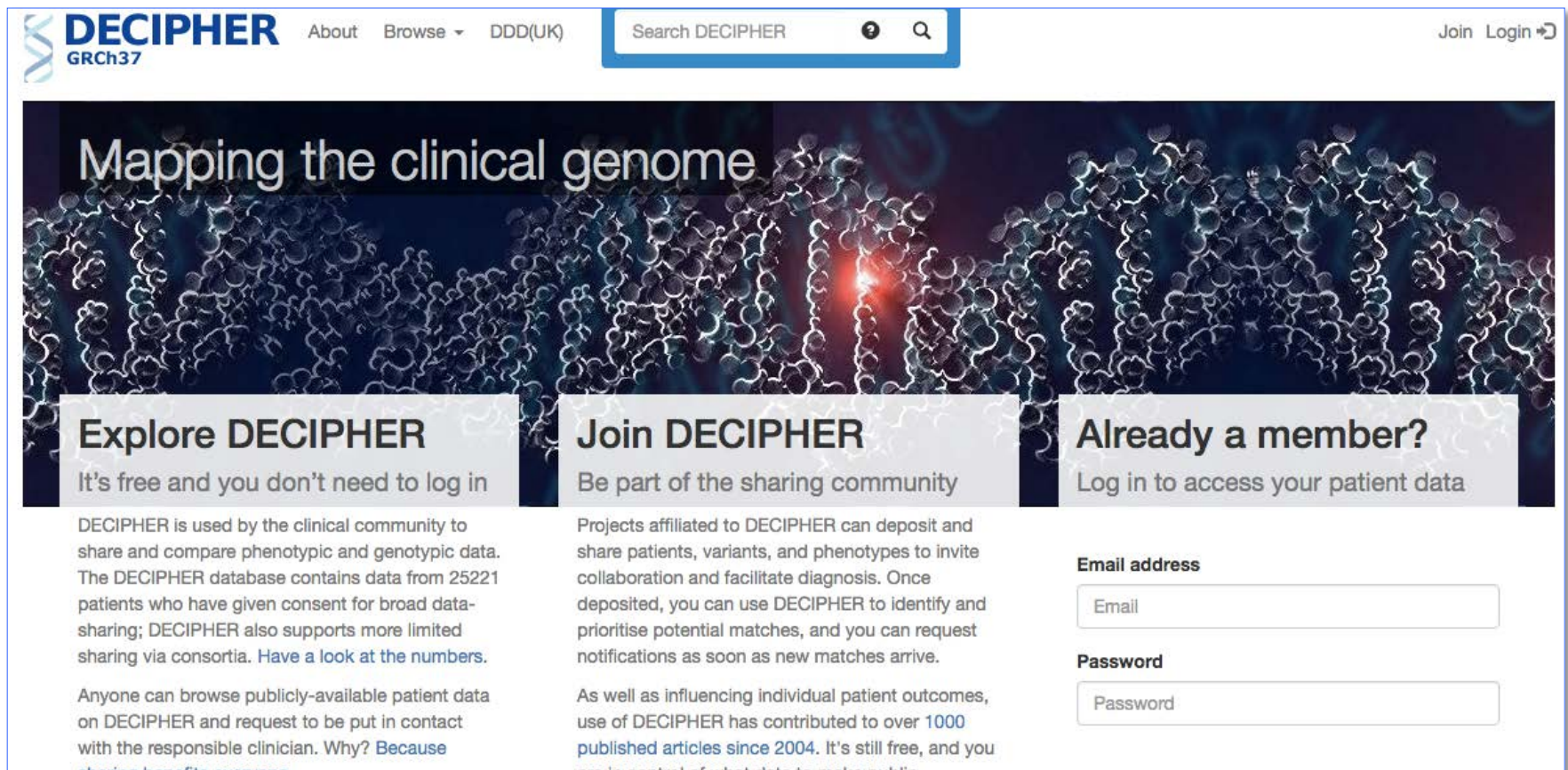
**This tool is provided for research use only. The BOADICEA software is at an early stage of development and is provided "as is" (ie. it is not error-free). BOADICEA is designed for research use only and is not designed for providing information on which to base clinical decisions. BOADICEA has not been approved for use by any regulatory authority.**

For any BOADICEA inquiry please contact Alex Cunningham ([apc40@medschl.cam.ac.uk](mailto:apc40@medschl.cam.ac.uk)) or Antonis Antoniou ([aca20@medschl.cam.ac.uk](mailto:aca20@medschl.cam.ac.uk)).

# DECIPHER

(Database of genomic variation and Phenotype in Humans using Ensembl Resources)

- The Deciphering Developmental Disorders (DDD) Study を元に構築されたデータベース (<https://decipher.sanger.ac.uk/>)



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## Mapping the clinical genome

### Explore DECIPHER

It's free and you don't need to log in

DECIPHER is used by the clinical community to share and compare phenotypic and genotypic data. The DECIPHER database contains data from 25221 patients who have given consent for broad data-sharing; DECIPHER also supports more limited sharing via consortia. [Have a look at the numbers.](#)

Anyone can browse publicly-available patient data on DECIPHER and request to be put in contact with the responsible clinician. Why? Because [sharing benefits everyone.](#)

### Join DECIPHER

Be part of the sharing community

Projects affiliated to DECIPHER can deposit and share patients, variants, and phenotypes to invite collaboration and facilitate diagnosis. Once deposited, you can use DECIPHER to identify and prioritise potential matches, and you can request notifications as soon as new matches arrive.

As well as influencing individual patient outcomes, use of DECIPHER has contributed to over 1000 published articles since 2004. It's still free, and you are in control of what data to make public.

### Already a member?

Log in to access your patient data

**Email address**

**Password**