

# PROBAND



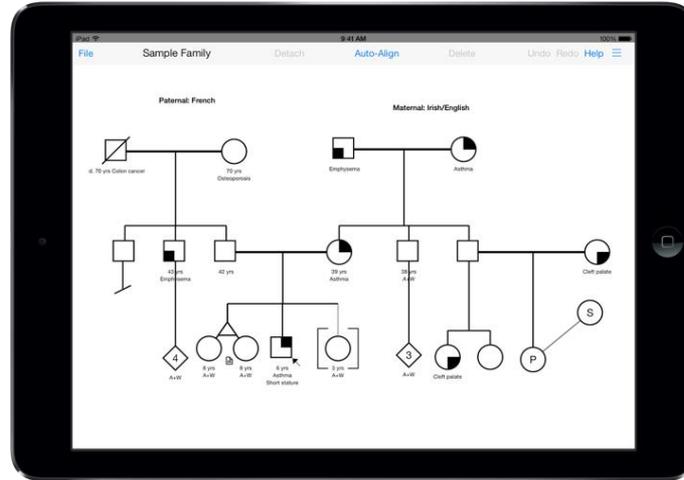
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# Origin

- Proband was created as part of the PediSeq Project (NHGRI 5U01HG006546-03) to help with structured **phenotype capture**
- One common requirement across disease areas was a detailed **family history**

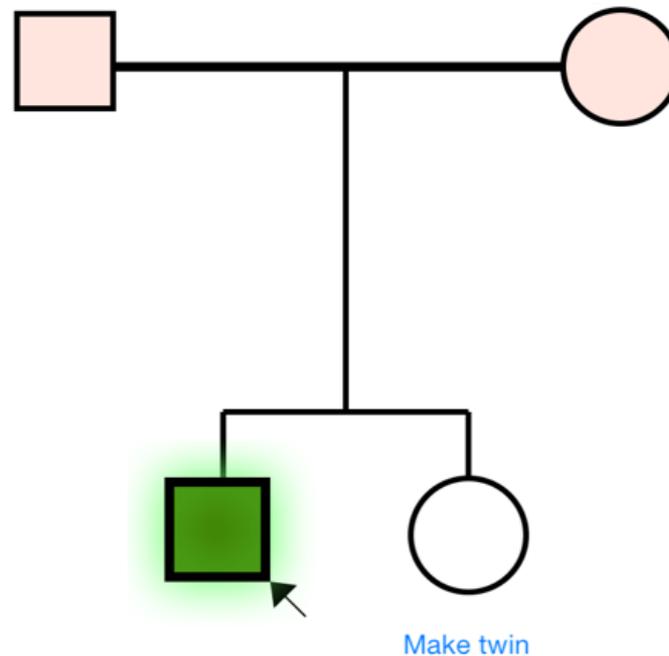
# PROBAND



- iPad only application, created for genetic counselors
- Intended to replace paper during an interview
- Captures structured, computable data
- Designed with input from genetic counselors at CHOP

# Inputs

- Proband uses the tablet interface to enable efficient data input

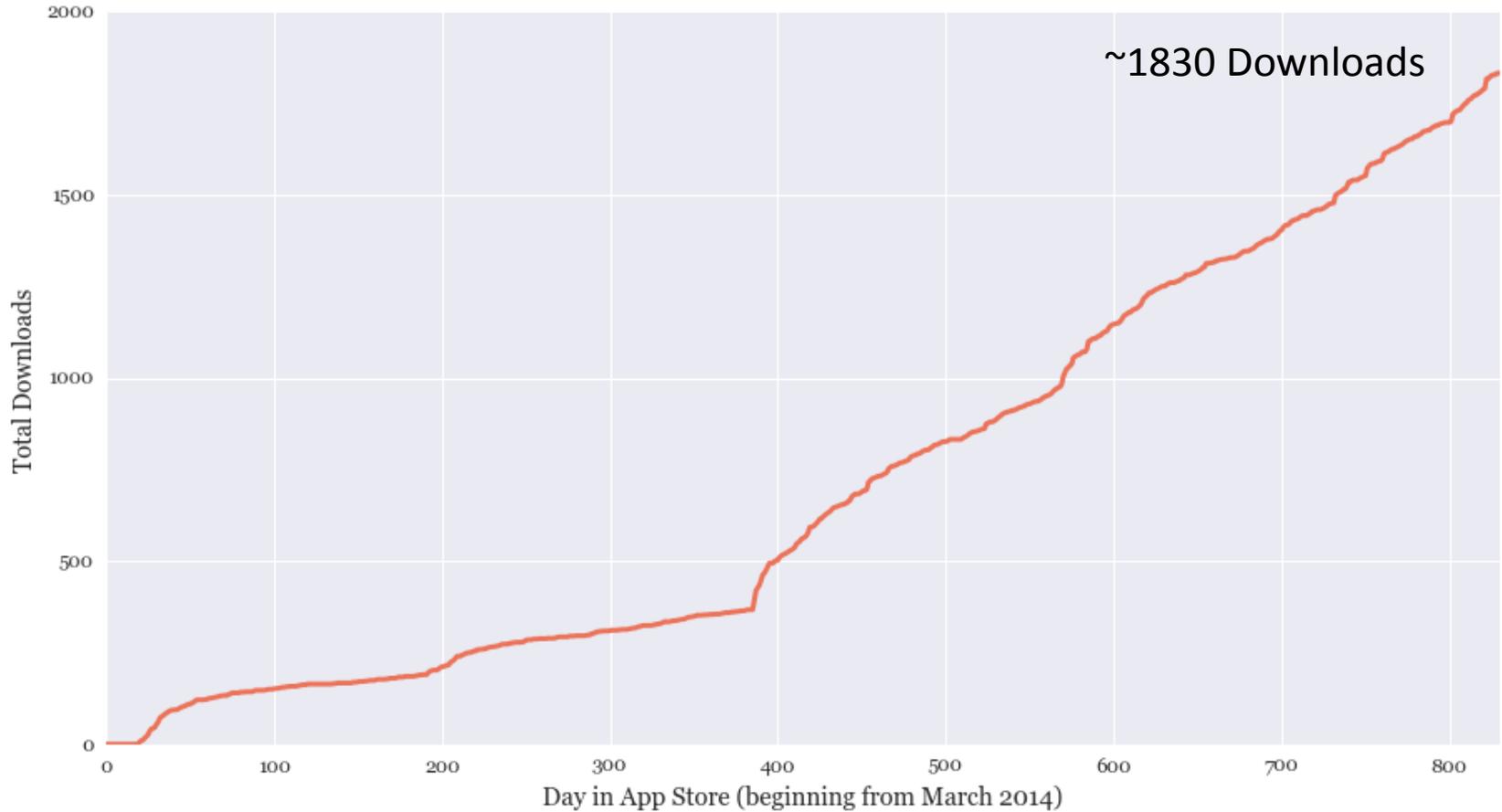


# Inputs

ICD10  
and  
HPO

The screenshot shows the 'testpedigree' application interface. At the top, there is a menu bar with options: File, testpedigree, Detach, Auto-Align, Delete, Undo, Redo, Help, and a hamburger menu icon. The main area displays a pedigree chart on the left with a highlighted individual. A pop-up form for this individual contains a 2x2 quadrant key (top-left: 4, top-right: 1, bottom-left: 3, bottom-right: 2) and a list of four disease input fields. The first field is 'Asthma' with a dropdown set to 'HPO' and a value of '6'. The second field is 'Scoliosis' with a dropdown set to 'ICD' and a value of '9'. The third field contains the text 'thyroi'. Below the form is a note: 'Note: Changing disease-quadrant key will affect entire pedigree, not only this person.' At the bottom of the application, a blue keyboard is visible with a search bar containing the text 'Thyroiditis HPO Thyroid adenoma HPO Thyroid agenesis HPO Thyroid carcinoma HPO Thyroid hypoplasia HPO Thyroid hyperplasia HPO'. A blue arrow points from the text 'ICD10 and HPO' to the search bar.

# Adoption



# Details

- Proband is free in the iPad App Store
- Exports to PDF, PNG and structured XML
- A server for backup, sharing and integration is currently in pilot phase and available under a software license agreement
- Contact Jeff Miller, [millerjm1@email.chop.edu](mailto:millerjm1@email.chop.edu)



# Poster Presentations

- **Incorporating Structured Pedigree Data into Genomic Medicine through a Pedigree and Family History iPad App**  
Jeffrey M. Miller, Michael J. Italia, Elizabeth T. Dechene, Alisha Wilkens, Mindy H. Li, Colleen E. Gaynor, Nancy B. Spinner, Ian D. Krantz, Jeffrey W. Pennington, Peter S. White  
Poster presented at American College of Medical Genetics Annual Clinical Genetics Meeting, March 25-29, 2014. Nashville, TN
- **Improving pedigree capture: Development and validation of an interview-optimized iPad app to eliminate the need for paper**  
Jeffrey M. Miller, Michael J. Italia, Elizabeth T. Dechene, Alisha Wilkens, Colleen E. Gaynor, Nancy B. Spinner, Ian D. Krantz, Jeffrey W. Pennington, Peter S. White  
Poster presented at the 64th Annual Meeting of The American Society of Human Genetics. Oct 18-22, 2014. San Diego, CA
- **Evolution of an mHealth Family Pedigree App**  
Jeffrey Miller, Donna Vito, Jeffrey Pennington  
Poster presented at AMIA Joints Summits. March 21-24, 2016. San Francisco, CA

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