A robust open science data platform for mechanistic disease discovery in Fanconi Anemia

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ABSTRACT

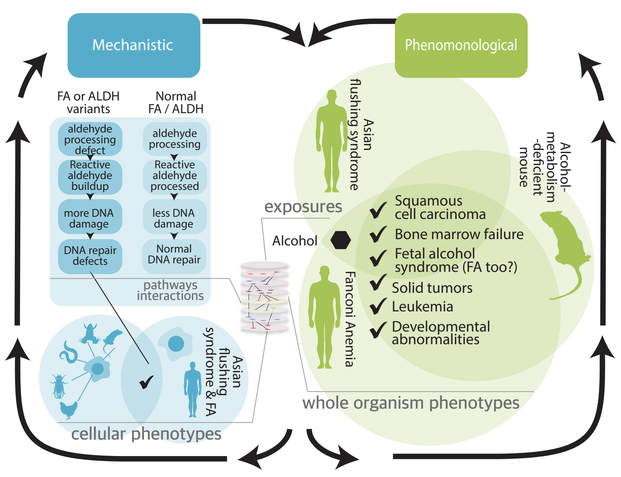
NEED
To address urgent unmet medical needs for rare diseases like Fanconi anemia, coordinated mining of data sets are needed to find relationships among molecular and cellular processes to illuminate potential drug targets. Unfortunately, data sources can be unshared, dispersed, incompatible, or not useful for inexpert users.

GOAL
Develop a new "Biomedical Data Translator" for the rare disease research community that integrates multiple types of existing data across the translational spectrum, including objective signs and symptoms of disease, drug effects and intervening types of biological data relevant to understanding pathophysiology to develop new mechanistic nosologies.

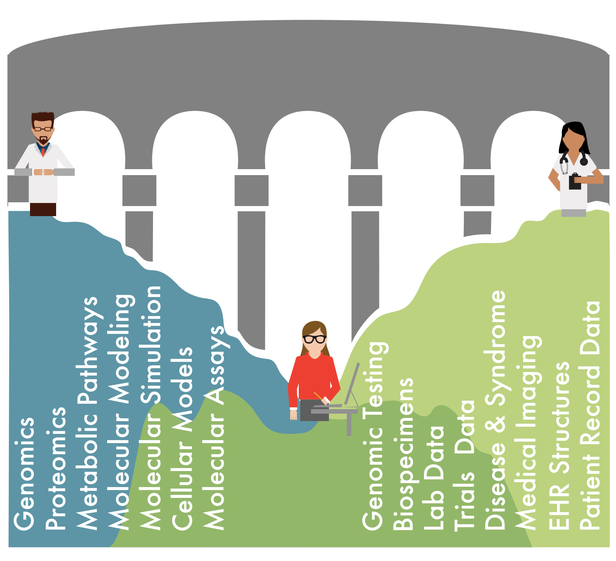
METHODS AND RESULTS
Fanconi anemia is one of the "demonstrator" projects for initial development and evaluation of the Translator. Analysis includes gene-environment interactions, synthetic clinical data, genetic variants, and insights from model organisms. The presentation will describe this innovative project and methods in detail, as well as initial results, with a focus on engaging the FA community in data sharing to advance this project.

TRANSLATIONAL IMPACT
We expect Translator will enable new insight into Fanconi anemia for improved diagnostics, prognosis, and treatment selection, and to support research by patients and advocates, basic scientists, drug developers, and clinical researchers.

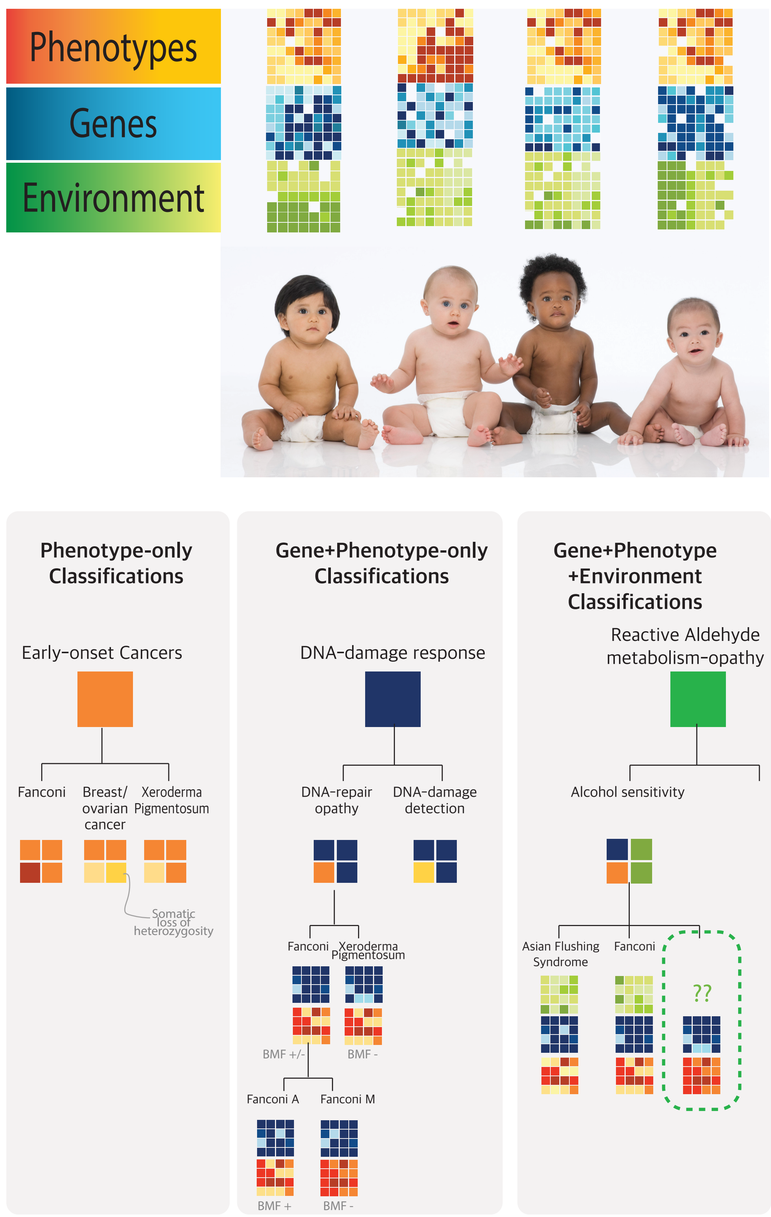
MECHANISTIC AND PHENOMONOLOGICAL BASIS



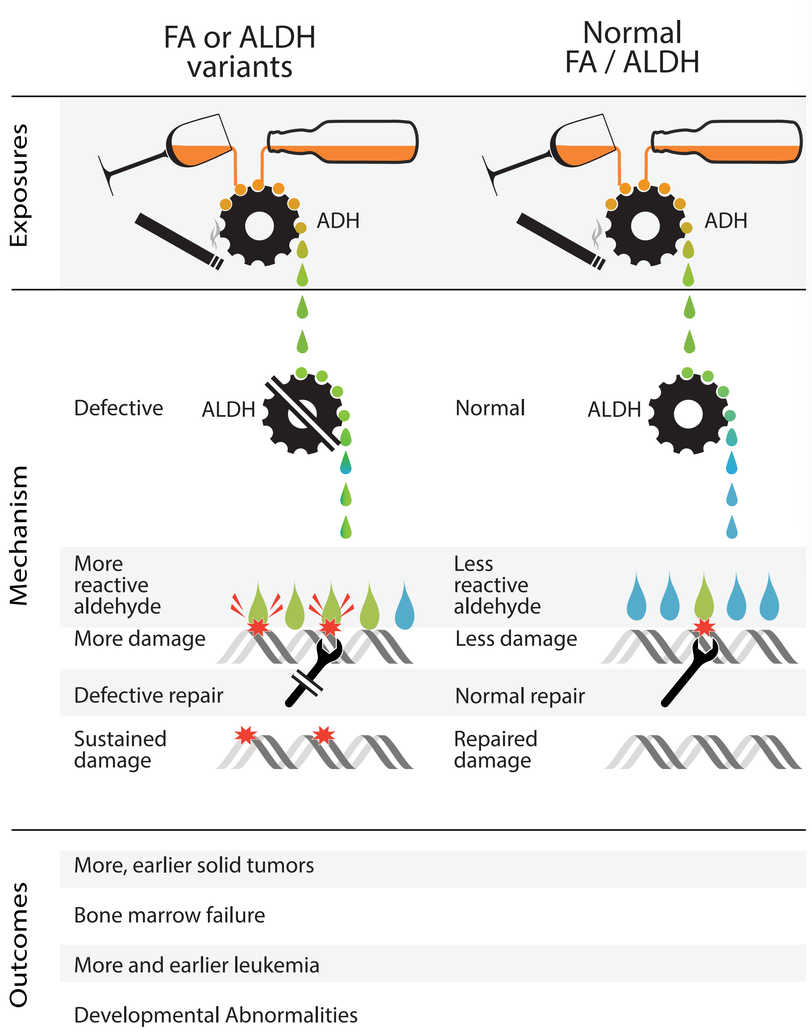
BRIDGING THE CHASM OF SEMANTIC DESPAIR



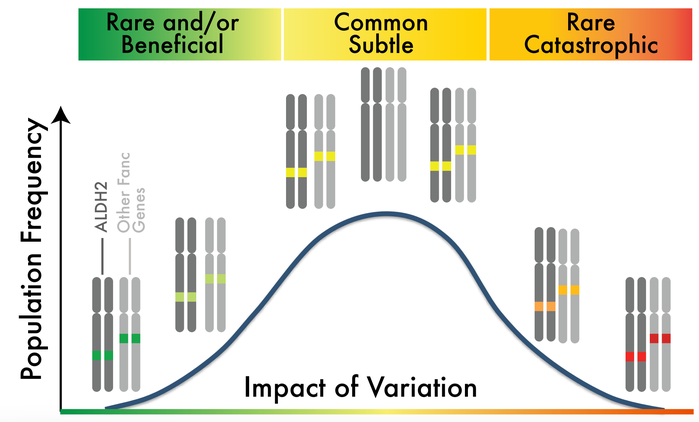
FANCONI ANEMIA: COMPLEX ETIOLOGY



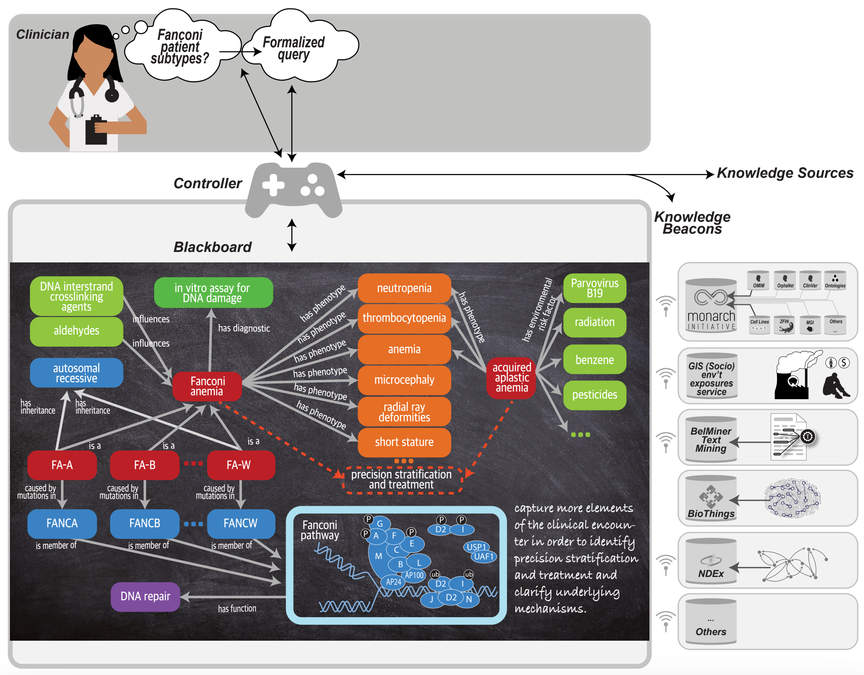
A FANCONI MECHANISM



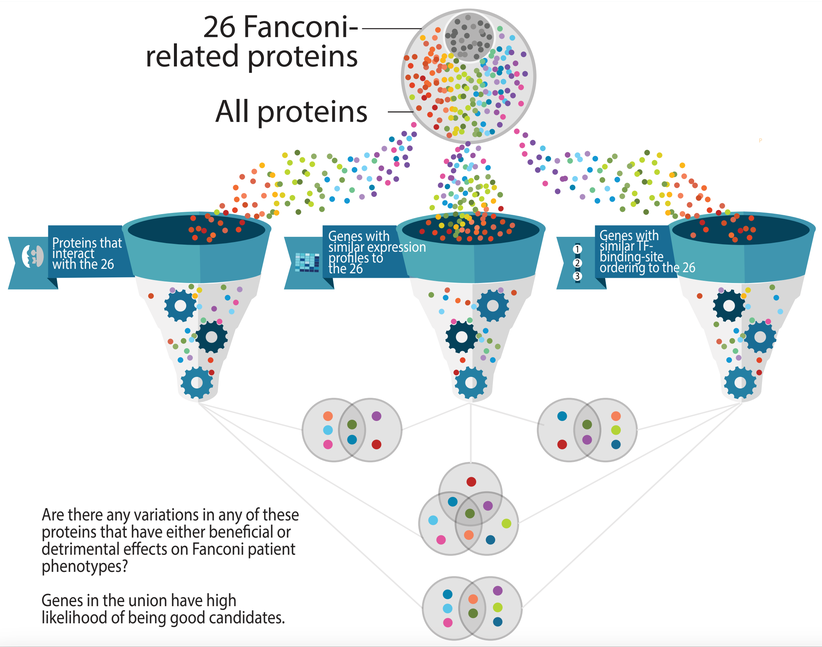
IMPACT OF VARATION IN FANCONI GENES



PROJECT APPROACH



QUERYING THE TRANSLATOR FOR FANCONI



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Technical Feasibility Assessment and Architecture Design
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