curadigm
Data to Insights

CASE STUDY

Genomic variants in autoinflammatory diseases: an association study

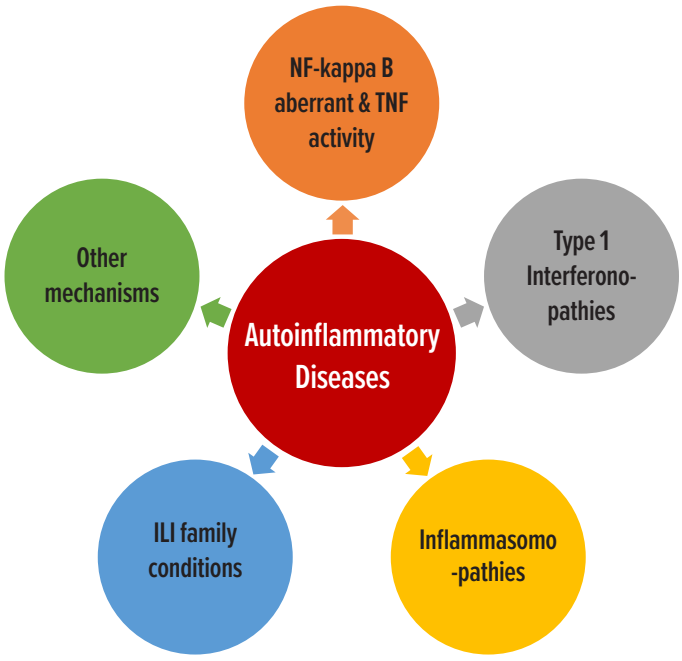
Objective

The customer was seeking to have a comprehensive coverage of literature based molecular information specific to auto-inflammatory disorders (AID), specific to genetic variants and genetic alternations, and develop a database of clinical genomic variants associated with autoinflammatory diseases (AI). In addition, the customer wanted to have a user-friendly dashboard to access the information by diversified users, primarily medical doctors associated with university hospitals.

Scope of the project

A list of 36 diseases related to AIDS across 4 groups was shared by the client.

The goal was to examine scientific publications and create a curated database of clinical genomic variants linked to autoinflammatory disorders (AID). Data that pertains to gene mutations, gene regulation, genotype-phenotype correlation, drug interventional studies, patient characteristics, experimental details, and detailed disease information including types and subtypes have been curated.

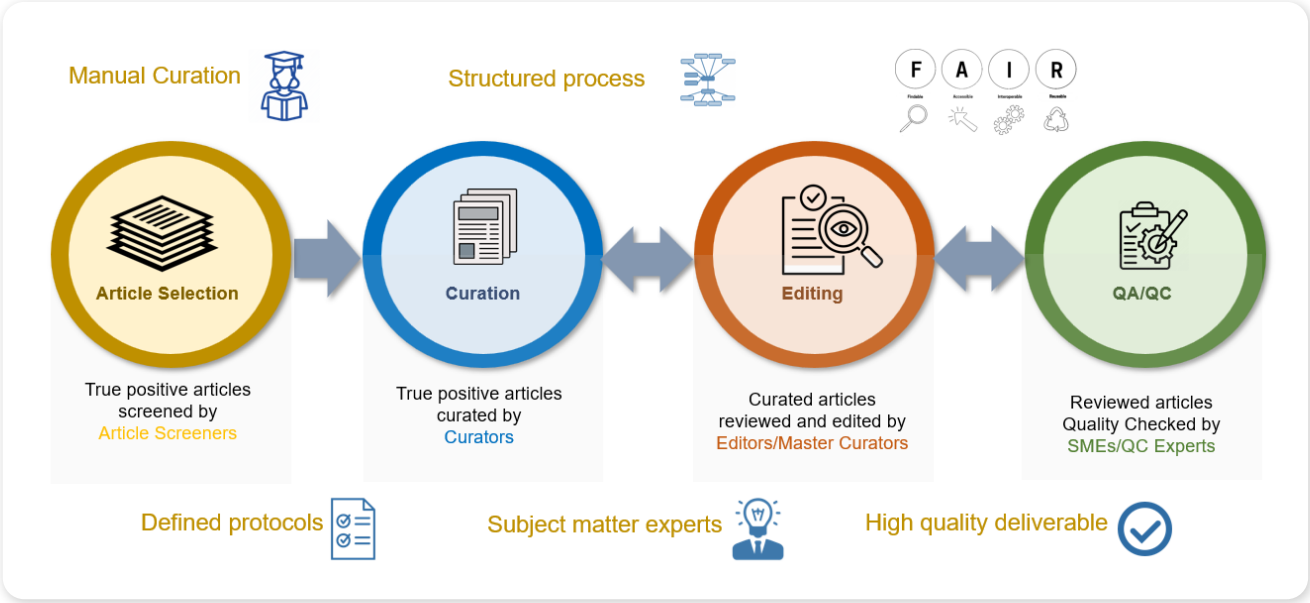


Search strategy

Curadigm utilized a variety of search methods to screen papers that pertain to autoinflammatory diseases and genetic variations that are linked to the disease. The use of search terminologies such as MeSH and other standard vocabularies was employed to identify articles. We had to continuously optimize search parameters as we progressed with curation.

Curation Process

We emphasize high quality deliverable through two level QC approach

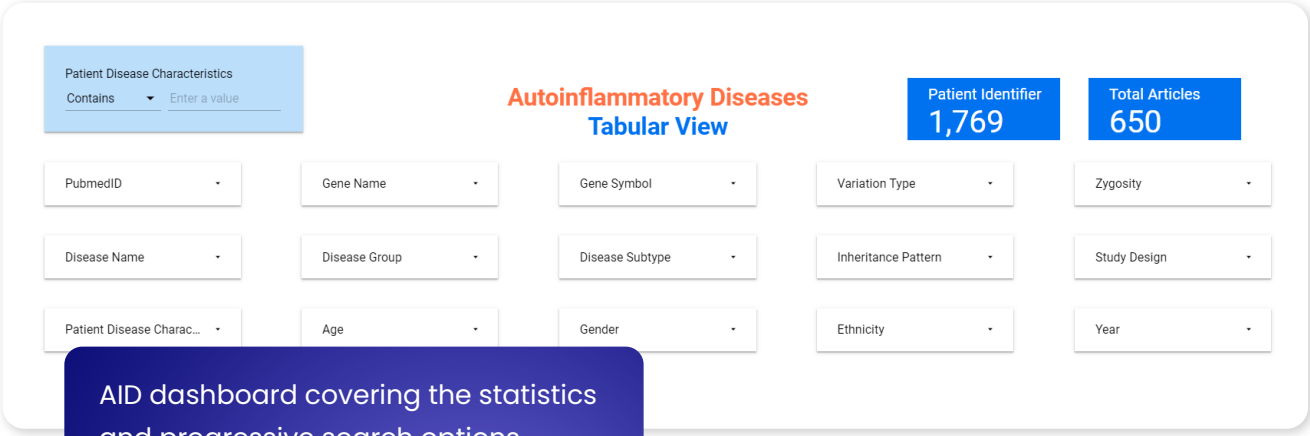


Results

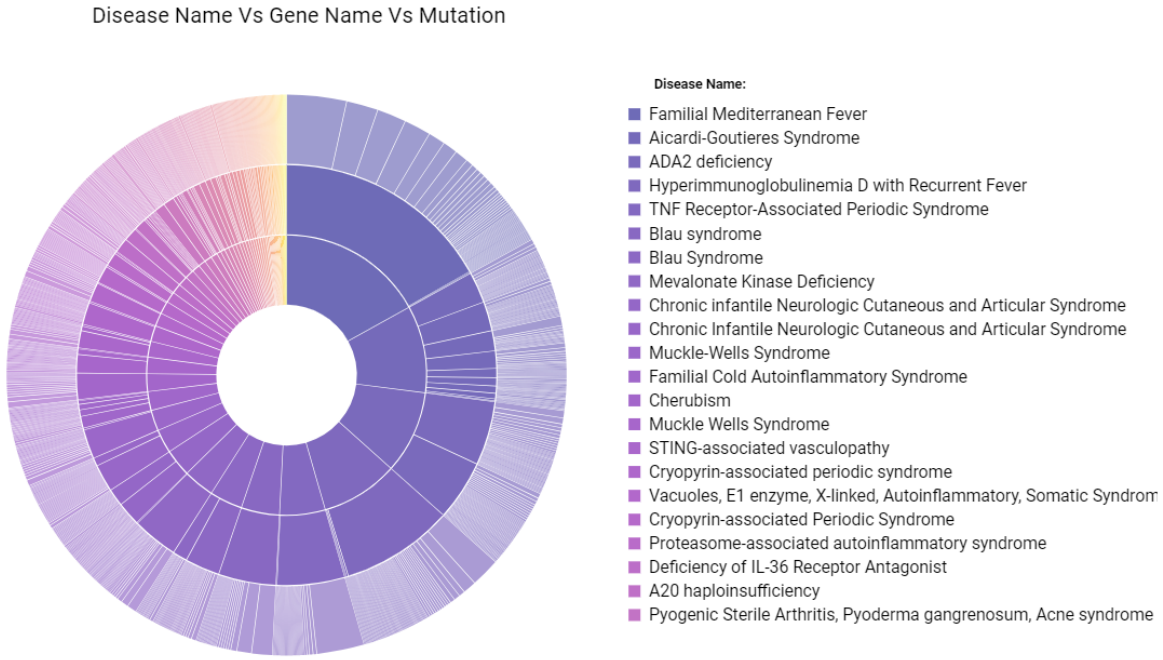
Curated information was delivered in the form of excel file, as well as searchable dashboard.

	PubmedID	Patient Identifier	Disease Name	Disease Group	Disease Subtype	Gene Name	Gene Symbol	Variation Type	Nucleotide HGVS	Amino Acid HGVS	Zygosity	Inheritance Pattern	Study Design	Patient Disease Characteristics
101.	30647181	30647181_P1	ADA2 deficiency	NF-kB and/or aberrant TNF activity	[NA]	ADA2	ADA2	Missense	NM_001282226.2:c.1078A>G	NP_001269155.1:p.Thr360Ala	[NA]	Autosomal recessive	Case study; Family study	Fever; Livedo reticularis- Trunk and Limbs; Inefficacy; Arthralgia; Chest pain; Abdominal pain; Gastrointestinal symptoms
102.	30647181	30647181_P1	ADA2 deficiency	NF-kB and/or aberrant TNF activity	[NA]	ADA2	ADA2	Deletion	NM_001282225.2:c.144del	NP_001269154.1:p.Arg49fs	[NA]	Autosomal recessive	Case study; Family study	Fever; Livedo reticularis- Trunk and Limbs; Inefficacy; Arthralgia; Chest pain; Abdominal pain; Gastrointestinal symptoms
103.	30647181	30647181_P3	ADA2 deficiency	NF-kB and/or aberrant TNF activity	[NA]	ADA2	ADA2	Missense	NM_001282226.2:c.1031C>T	NP_001269155.1:p.Pro344Leu	Compound Heterozygous	Autosomal recessive	Case study; Family study	Recurrent fever; Arthralgia; Gastrointestinal symptoms; Arthritis; Livedo reticularis- Limbs; Seizures; Neurosensorial hearing loss; Hypertension; Psoriasis; Episcleritis; Behavioral disorder

Fewer important data fields are presented above



AID dashboard covering the statistics and progressive search options



Integrated Sunburst view to search more specific datasets

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