

## CASE STUDY

# Genes and Hallmarks in metabolic diseases: an association study

### **Objective**

The customer was seeking to develop a concept of hallmark-mediated disease mechanisms throughout the entire metabolic spectrum, with a specific focus on obesity, and identify the hallmarks of obesity and gather gene sets and molecular alterations related to them, along with supporting evidence.

### **Curation process**

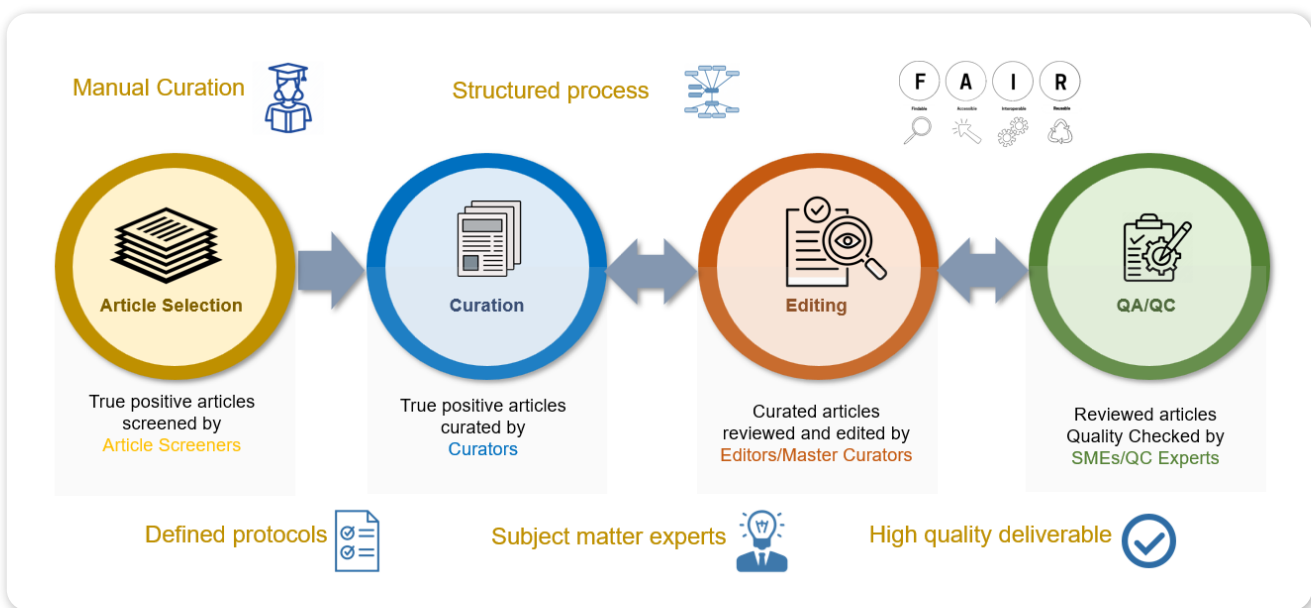
Article selection and identification of relevant articles that support the molecular level association with obesity were the initial steps of the curation process. The next step was to manually curate full-text articles. Structured format was used to capture information about obesity, including differential gene regulation, protein interactions, altered pathways, and phenotypes. To link genes and altered mechanisms, the obesity hallmark ontology was developed.

## Article search strategy

Curadigm utilized multiple search strategies to screen papers that pertain to obesity and molecular mechanisms associated with the disease. To identify articles, search terminologies such as MeSH and other standard vocabularies were used. As we progressed with curation and defining the hallmarks, it was necessary to continuously optimize search parameters.

## Curation Process

We emphasize high quality deliverable through two level QC approach



## Results

The information that was curated was presented in an excel file and a searchable dashboard.

PubMed	Disease	From Node	Molecular Effect	Degree of Alterati	To Node	Molecul	Degree of /	Phenoty	Phenoty	Hallmark Level 1	Hallmark Level 2	BMI Details	Study Type	
32079412	Obesity	Leptin	Level	Increased	hsCRP	Level	Negative assoc	Inflammatio	Associated	Immune	Synovium Inflammatio	Mean: 30.5 (6)	Clinical	
19837927	Obesity	COL6A3	Expression	Increased	MIP-1 alpha	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 33.5 +/-	Clinical	
31652924	Obesity	Vitamin D rece	Expression	Increased	IL-1-beta	Expression	Positive assoc	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 46.85 A	Clinical	
31652924	Obesity	Vitamin D rece	Expression	Increased	IL-6	Expression	Positive assoc	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 46.85 A	Clinical	
31652924	Obesity	Vitamin D rece	Expression	Increased	IL-8	Expression	Positive assoc	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 46.85 A	Clinical	
32079412	Obesity	Leptin	Level	Increased	hsCRP	Level	Positive assoc	Inflammatio	Associated	Immune	Alteration of metabo	Changes in Serum infl	Mean: 30.5 (6)	Clinical
32079412	Obesity	Leptin	Level	Increased	IL-6	Level	Positive assoc	Inflammatio	Associated	Immune	Alteration of metabo	Changes in Serum infl	Mean: 30.5 (6)	Clinical
32079412	Obesity	Leptin	Level	Increased	TNF-alpha	Level	Negative assoc	Inflammatio	Associated	Immune	Alteration of metabo	Changes in Serum infl	Mean: 30.5 (6)	Clinical
32079412	Obesity	Leptin	Level	Increased	TNF-alpha	Level	Positive assoc	Inflammatio	Associated	Immune	Synovium Inflammatio	Mean: 30.5 (6)	Clinical	
32079412	Obesity	Leptin	Level	Increased	IL-6	Level	Positive assoc	Inflammatio	Associated	Immune	Synovium Inflammatio	Mean: 30.5 (6)	Clinical	
19837927	Obesity	COL6A3	Expression	Increased	MCP1 alph	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 33.5 +/-	Clinical	
19837927	Obesity	COL6A3	Expression	Increased	CD163	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 33.5 +/-	Clinical	
23179203	Obesity	STEAP4	Expression	Increased	CD68	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	
23179203	Obesity	STEAP4	Expression	Increased	MMP2	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	
23179203	Obesity	STEAP4	Expression	Increased	MMP9	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	
23179203	Obesity	STEAP4	Expression	Increased	TNF-alpha	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	
23179203	Obesity	STEAP4	Expression	Increased	SPP1	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	
23179203	Obesity	NGAL	Expression	Increased	CD68	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	
23179203	Obesity	NGAL	Expression	Increased	MMP2	Expression	Increased	Inflammatio	Associated	Immune	Adipose tissue & meta	Mean: 45.4 A	Clinical	

Fewer important data fields are presented above

## Obesity dashboard view

**Delivery Status**

Article ID -

Hallmark Level 2 -

Biomarker -

From Node Name -

Study Type: Clinical (1) -

Disease Name -

**Hallmark Level 2 vs PubMed ID**

From Node (Gene Symbol) -

To Node Name -

To Node (Gene Symbol) -

Patient Sample Size (Case) -

Year -

Phenotype -

Hallmark Level 2	Unique PubMedID	No. of Records
1. ALP	2	3
2. ALT	21	33
3. AST	14	24
4. Acute insulin respo...	4	8
5. Adipocyte hyper- & ...	12	45
6. Adipocyte hypertro...	8	24
7. Adipocyte hypotrop...	3	85
8. Adipogenesis & adi...	8	34
9. Adipose tissue & m...	41	555
10. Adipose tissue cell...	2	6
11. Adipose tissue hyp...	4	19
12. Adipose tissue ma...	10	40
13. Apolipoprotein A-I	1	1
14. Apolipoprotein B (A...	5	7
15. Arginine in plasma	1	3
16. Arginine in plasma: ...	1	1
17. Arginine in plasma: ...	1	1
18. Beiging of white ad...	2	6
19. Biomechanics of th...	3	73
20. Blood pressure	2	5

**Hallmark Level 2:**

- Whole body fat distribution (across all tiss
- [NA]
- Loss of articular cartilage
- Adipose tissue & metabolic inflammation
- HOMA-IR
- Triglycerides (triaclycerol/TG) (includin
- Joint physical function
- Inflammation
- High density lipoproteins (HDL)
- Joint pain
- Hepatic steatosis
- Glucose levels
- De novo lipogenesis (DNL)
- Oxidative stress/Lipid peroxidation
- Small dense low density lipoprotein (LDL)
- Insulin levels
- Cholesterol
- Fasting insulin
- Liver fibrosis
- Fasting glucose
- Changes in Serum inflammatory mediator:

The Sunburst view has been integrated to search for more specific datasets

For more information, contact us at [info@curadigmdata.com](mailto:info@curadigmdata.com)



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