Rare Connect

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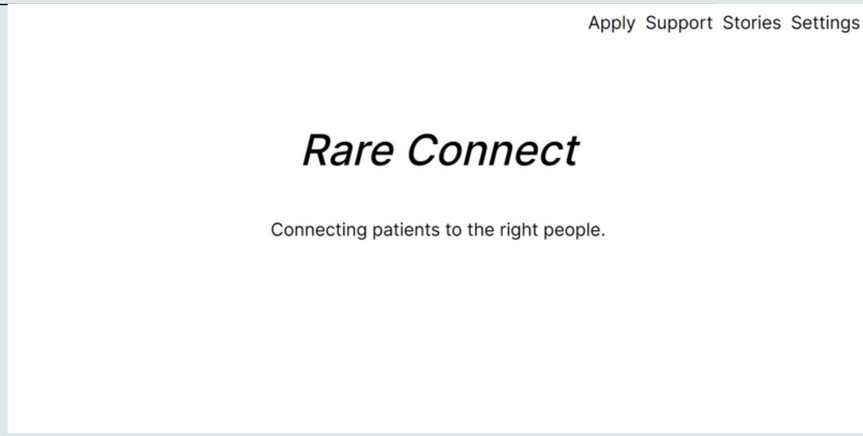
Problem You're Tackling

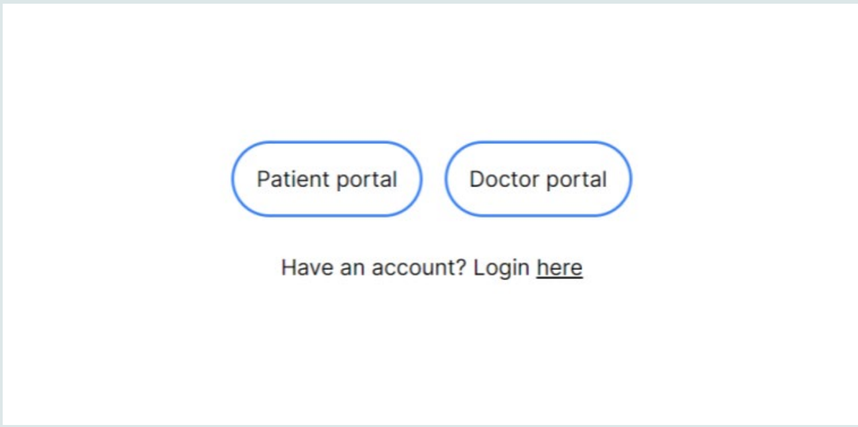
Misdiagnosis and Access to Information

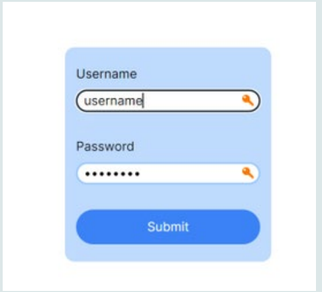
- Challenge: Individuals with rare diseases often struggle with getting an accurate diagnosis, leading to delays in treatment and increased psychological stress.

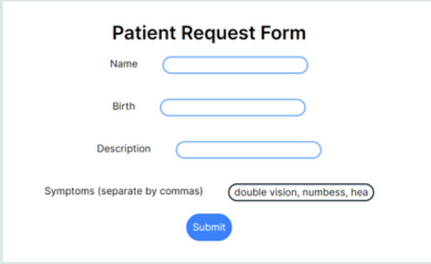
Isolation and Lack of Support

- Challenge: The rarity of these conditions can lead to isolation and a lack of understanding from the broader community.









Relevant Literature

- Websites
  - HIPAA
  - https://hodassoc.org
  - https://www.foxp1.org/characteristics
  - https://usp7.org/signs-and-symptoms
  - https://www.curecmt4j.org/cmt4j/
  - https://alportsyndrome.org/about-alport-syndrome/diagnosis/
  - https://dup15q.org/understanding-dup15q-syndrome/

Data Worked With

- Identified from rare disease websites
  - Symptoms
  - Demographics (has some weight but does not rule out minority populations)

1	Disease name	Common Symptoms	Less Common Symptoms	Age	Biological Gender	Website	Diagnosis Criteria Available
2	HOD	Balance/Gait Unsteadiness, Nystagmus, Double Vision, Fatigue, Myoclonus, Palatal Tremor, Ataxia/Poor, Coordination, Headache, Numbness, Pins and Needles feelings that don't resolve quickly	Spasticity, Difficulty Swallowing, Slurred Speech, Tinnitus,	-	-	https://hodassoc.org/	-
3	FoxP1	Delays in early motor and language milestones, Mild-to-severe intellectual deficits, Speech and language impairment in all individuals regardless of cognitive ability, behaviour abnormalities, autism spectrum disorder or autistic features, attention-deficit/hyperactivity disorder, anxiety, repetitive behaviors, sleep disturbances, and sensory symptoms	(contributing to speech and feeding difficulties), Refractive errors, Strabismus,	-	-	https://www.foxp1.org	No
4	USP7	Dysmorphic Facial, Abnormal MRI, Hypotonia, Eye Anomalies (esotropia, myopia, strabismus, nystagmus), Feeding Problems (including need for special feeding technique), GERD, Seizures, Neonatal Hypotonia, Hypogonadism, Asthma, Abnormal Gait, Difficulty Gaining Weight - Sleep Apnea/Sleep Disturbance, Chronic Constipation, Short Stature, Scoliosis or Kyphosis, Neonatal Poor Suck, Contractures, Small Hands, Small Feet, Excessive Weight Gain, Chronic Diarrhea, Decreased Fetal Movement, Hip Dysplasia, Hearing Difficulties, Speech Delay (some nonverbal), Developmental Delay/Intellectual Disability, Behavioral Anomalies (aggressive behavior, temper tantrums, impulsivity, compulsivity, stubbornness, manipulative behavior), Autism Spectrum Disorder, ADHD, Skin Picking	-	-	-	https://usp7.org	-

Psuedo Database

Methods

Artificial intelligence

- Python ML
- CNN’s

Backend

- Python

Frontend

- Typescript

Results

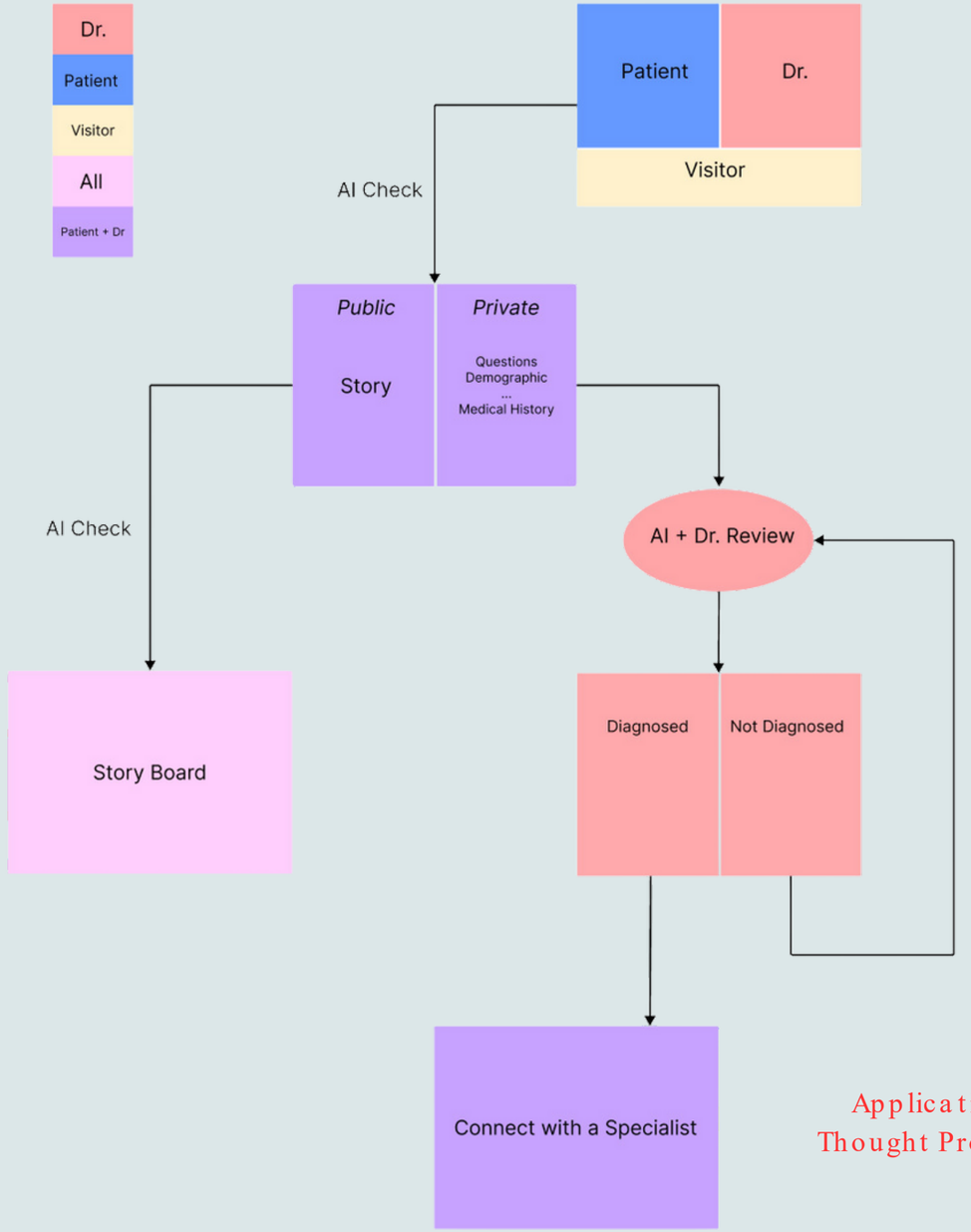
- Created a conceptual design of the platform
- Mock algorithm with sample data set to model AI


```
{"symptoms": ["double vision", "cough", "headache", "numbness", "fatigue", "seizures"]}
```


Symptoms


```
{
  "HOD": 4,
  "Behcet's Disease": 4,
  "USP7": 1
}
```


Expected Output



Application Thought Process

Discussion

- Limitations
  - Funding
  - Must be compliant with HIPAA
  - Outreach
- What's been done
  - Doctor Patient Online Chatting
  - https://compliantchatgpt.com/
- Why it's different
  - Focused on the Rare Disease space

Challenge and Future Directions

- Implementation
- Funding → getting sponsors or backing through companies or investors
- HIPAA compliance
- Making the platform a key aspect of the rare disease community