# The Greenwood Genetic Center: A Privileged Institution

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JC Self Research Institute
Greenwood Genetic Center



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# **Greenwood Genetic Center Origins - 1974**







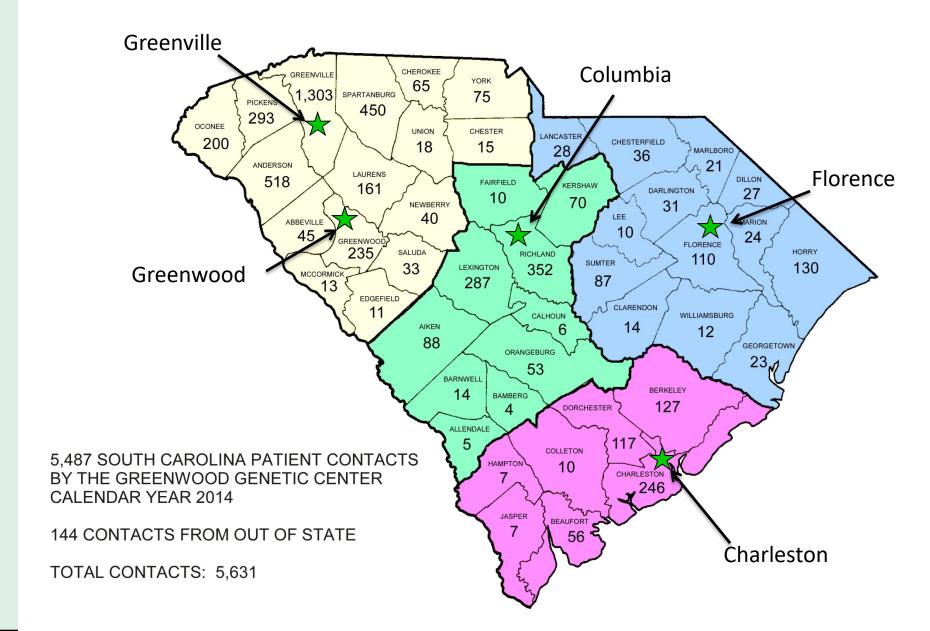
Private, non-profit institute

#### **Greenwood Genetic Center**

#### Mission:

- Provide clinical genetic services and diagnostic testing
- Develop educational programs and materials
- Conduct research in the field of medical genetics

### **Genetic Clinics**



# The Focus of the Greenwood Genetic Center

**Intellectual Disabilities** 

**Birth Defects** 

**Autism** 

# JC Self Research Institute of Human Genetics

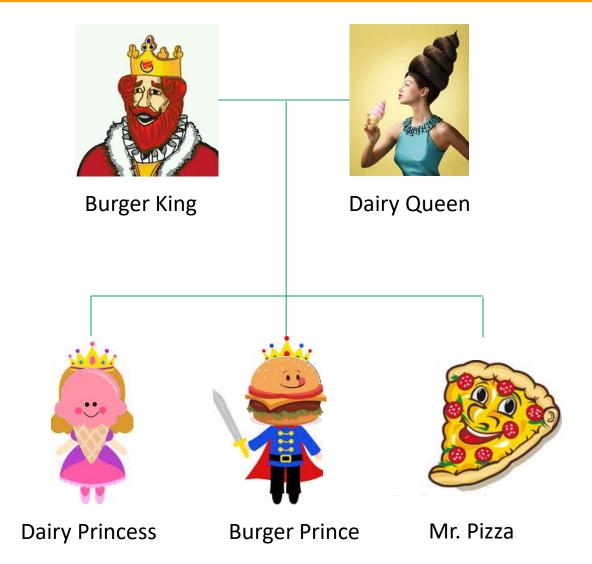
#### Research Division of the Greenwood Genetic Center

#### **Research Teams**

- Mental Retardation
- Autism
- Limb Malformations
- NTD / Birth Defects



## **That Recessive Gene**



#### **XLID**

Identified >28 XLID genes

Enrolled >700 families

Associated with numerous rare syndrome support groups

#### **NTD**

SC Birth Defects Prevention Program – 1991

Decreased rated of NTDs by >60%

Accounts for 70 healthy babies/year

### **Autism**

South Carolina Autism Project (SCAP): 1996
200 Probands

2<sup>nd</sup> Cohort ASD (2014): 100 Probands

3<sup>rd</sup> Cohort ASD (2015): 100 Probands

## **Capabilities**

- Next Generation Sequencing
- Microarray Analyses
- Biochemical Analysis
- Molecular Genetic Studies

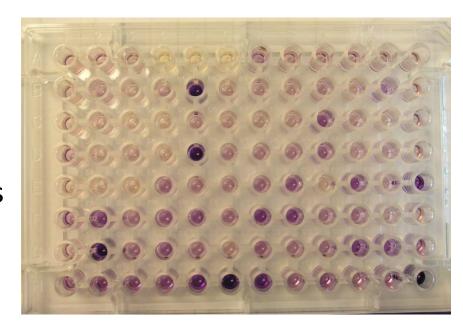
### **Collaborations**

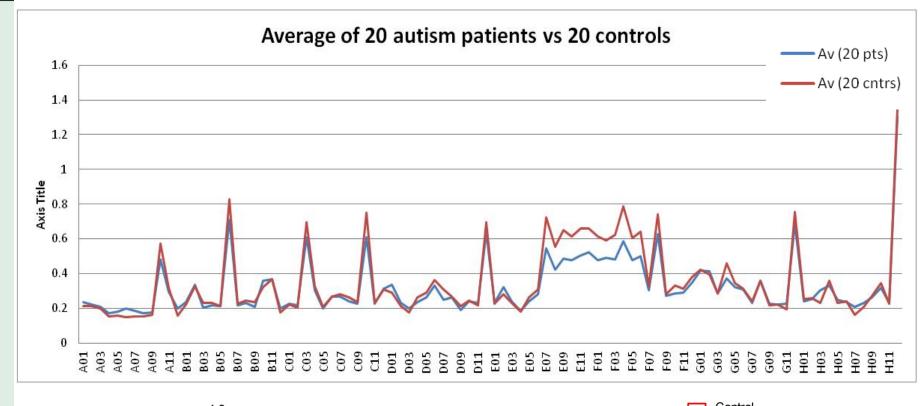
Functional Studies
 Electrophysiology
 Cell biology
 Animal models

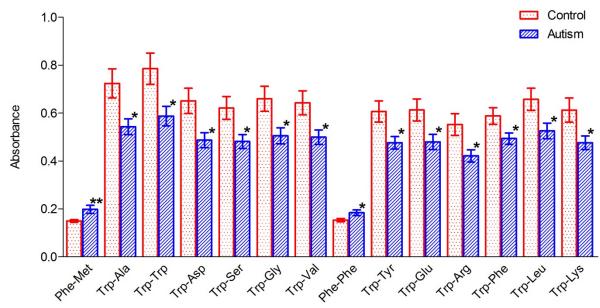
Proteomic Analyses

## **Biolog PMM Plates**

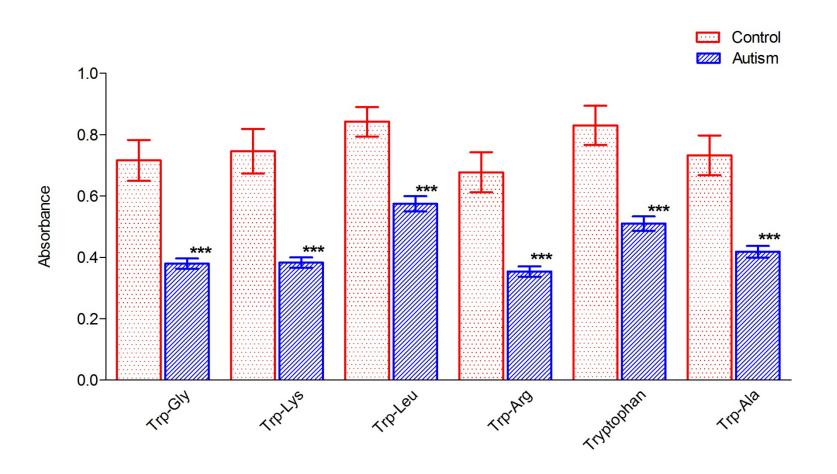
- 50 μl/well of cell suspension (20,000 cells/well)
- Incubate 40-48 hours, add Redox Dye Mix (tetrazolium) and incubate for 24 hours
- Plate substrates:
- Carbon energy sources (sugars, polysaccharides, nucleotides, and carboxylic acids)
- 2-4. Amino acids and dipeptides
- 5. lons
- 6-8. Hormones and metabolic effectors

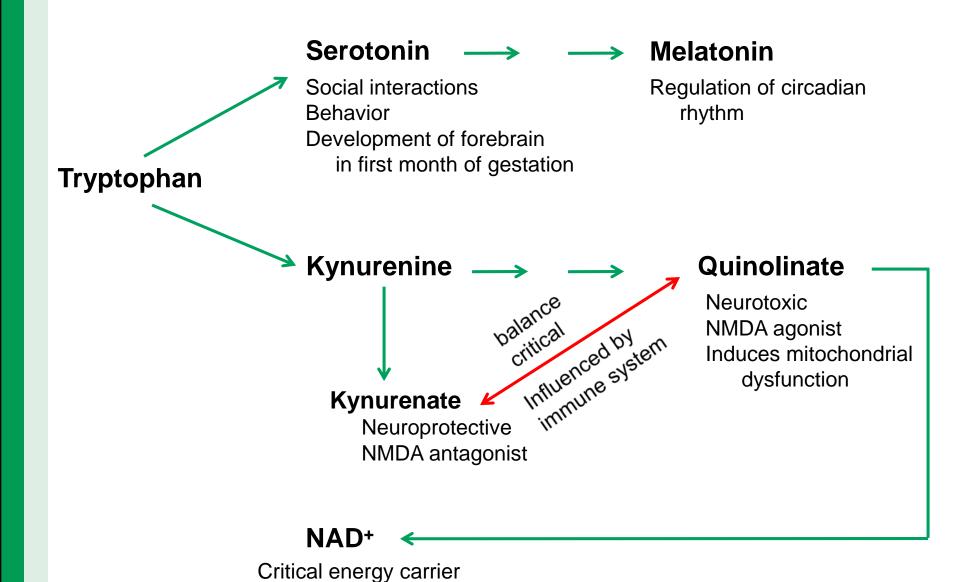






## 50 ASD patients vs. 50 controls





for electron transport

## **Screening Test**

#### With All Tryp

Sample	Correct	Error	
Autism	46	4	50
Control	33	17	50

Sensitivity = 92%

Specificity =

**Specificity = 66%** Bagging

With All Tryp and PM5				
Sample	Correct	Error		
Autism	46	4	50	
Control	46	4	50	
Sensitivity =	92%		_	

92%

Bagging

### **Greenwood Genetic Center**



It's in our genes