One Test, A Multitude of Rare Disease Diagnoses

Rady Children's Institute Genomic Medicine

- As of **October 2020**, RCIGM has reported a confirmed genetic diagnoses in 630 children. Of those 630 children, **540 were unique one gene-disease associations.** The others were recurrent genetic diagnoses seen in ≥2 patients.
- WGS of unselected patient populations (NICU, PICU) has revealed **expanded phenotypes** for previously well-characterized genetic conditions and highlighted the potential for WGS for **diagnosis of rare disease**

	Global Prevalence[+]	RCHSD Prevalence*	RCIGM Rare Disease Prevalence
Kabuki Syndrome	1-9 / 100,000	1 / 56,000	**1 / 140**
CHARGE Syndrome	1-9 / 100,000	1 / 56,000	**1 / 170**
KCNQ2-Related Epileptic Encephalopathy	<1 / 1,000,000	1 / 29,000[#]	**1 / 250**

[+] According to Orphanet database
* Based on unrefined/preliminary EMR search for primary condition or ICD-10
[#] RCHSD prevalence for "epileptic encephalopathy"