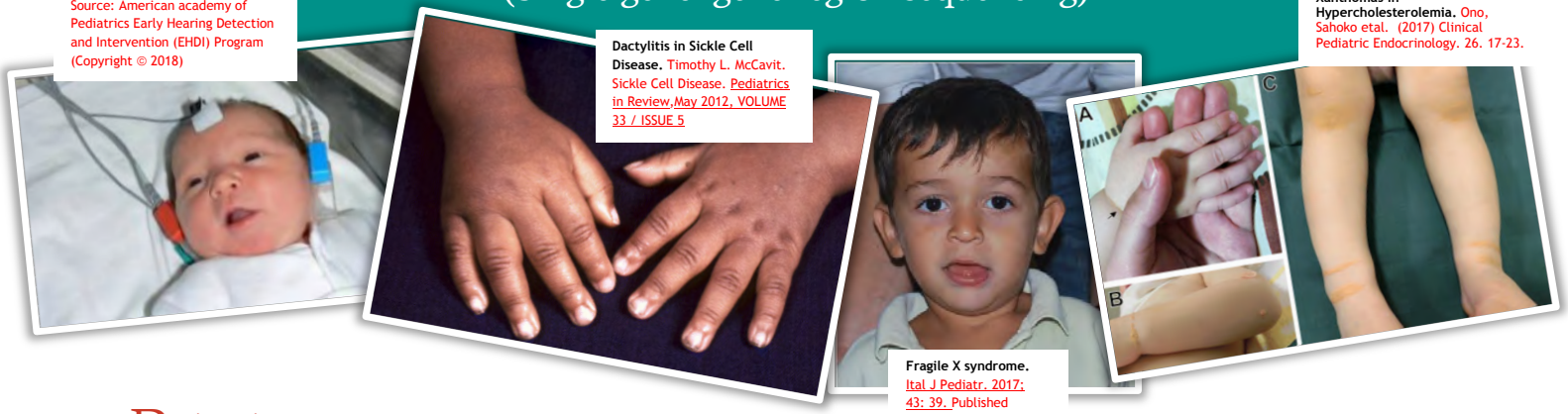
Targeted Gene Sequencing

(Single gene/gene region sequencing)



Newborn Hearing Screen. Source: American academy of Pediatrics Early Hearing Detection and Intervention (EHDI) Program (Copyright © 2018)

Dactylitis in Sickle Cell Disease. Timothy L. McCavit. Sickle Cell Disease. Pediatrics in Review,May 2012, VOLUME 33 / ISSUE 5

Fragile X syndrome. Ital J Pediatr. 2017; 43: 39. Published online 2017 Apr 19. doi: 10.1186/s13052-017-0355-y

Xanthomas in Hypercholesterolemia. Ono, Sahoko etal. (2017) Clinical Pediatric Endocrinology. 26. 17-23.

Detects:

- Gene mutation in single gene

Advantages:

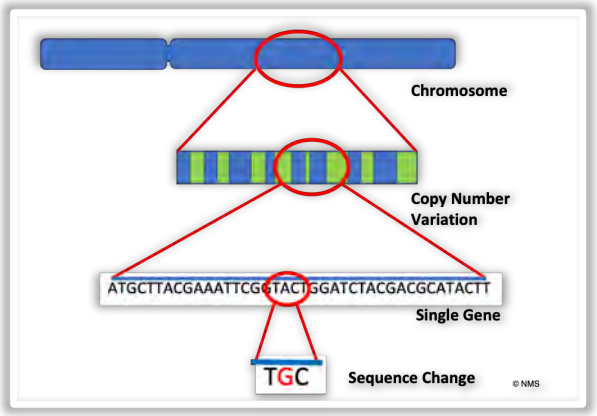
- Cost effective
- High fidelity results due to high depth of sequencing (~500X)

Limitations:

- Evaluation limited to a specific gene/gene region

Clinical Applications

- High index of suspicion for specific genetic condition with only 1 causative gene



Targeted Gene/Gene Region Sequencing

Condition	Gene (Chr. Location)	Mode of Inheritance
Congenital Deafness (nonsyndromic)	Connexin 26 (13q11)	Recessive
Tay-Sachs	hexosaminidase A (15q23)	Recessive
Familial hypercholesterolemia	LDL receptor (19p13)	Dominant
Sickle cell anemia	Beta-globin (11p15)	Recessive
Duchenne muscular dystrophy	Dystrophin (Xq21)	X-linked Recessive
Cystic Fibrosis	CFTR (7q31)	Recessive
Hemochromatosis	HFE (6p21)	Recessive
Huntington disease	Huntington (4p16)	Dominant
Fragile X syndrome	*FMR1* (Xq27.3)	X-linked Dominant

Modified from: Genetic Alliance; District of Columbia Department of Health. Understanding Genetics: A District of Columbia Guide for Patients and Health Professionals. Washington (DC): Genetic Alliance; 2010 Feb 17. Appendix G, Single-Gene Disorders. Available from: https://www.ncbi.nlm.nih.gov/books/NBK132154/