

HoFH WEBINAR TRILOGY

Advancing Knowledge, Care, and Community

Finding HoFH From the Very Beginning

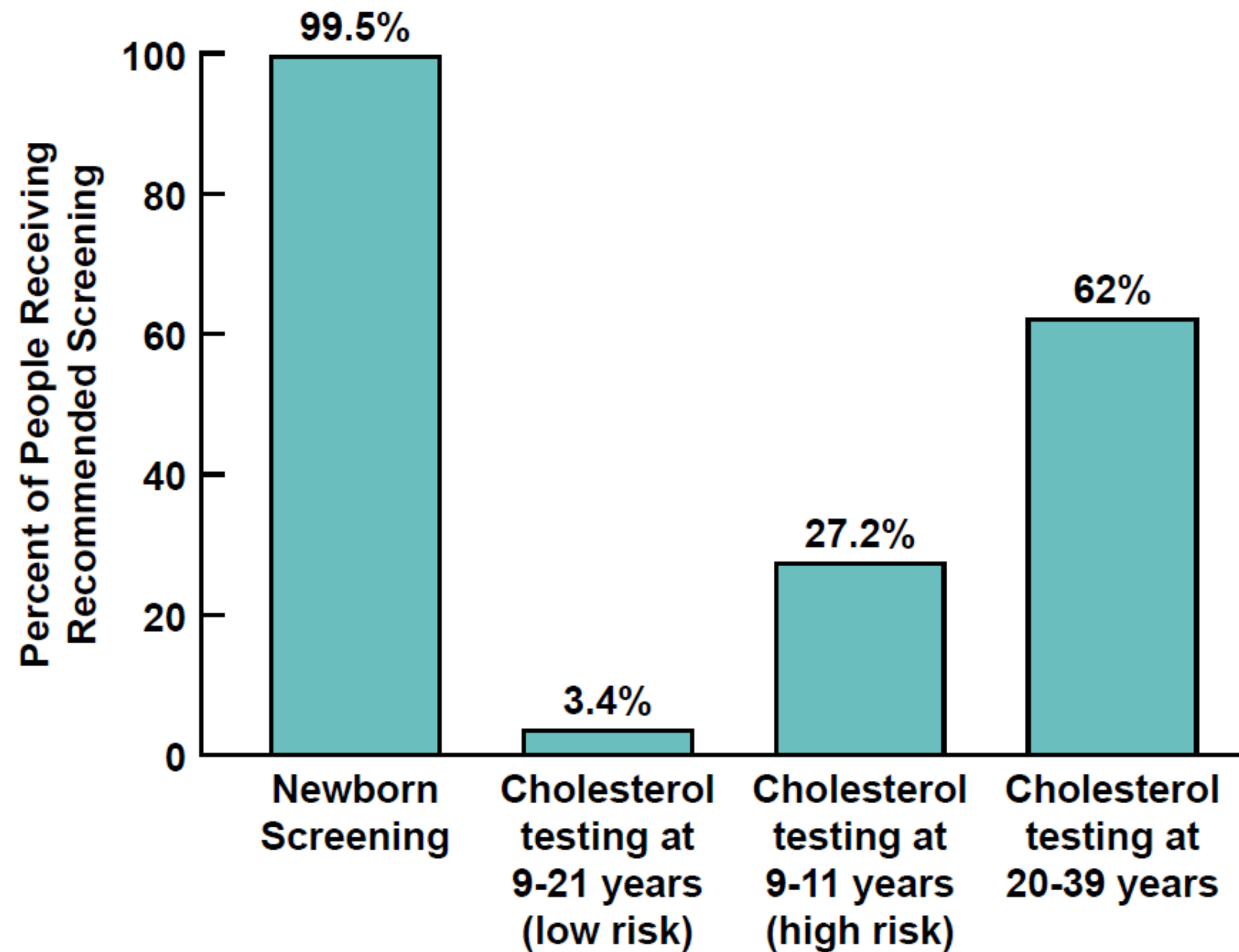
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Episode I. Starting Early: **Changing the Future of HoFH**



The Problem: People don't get their cholesterol checked.



- When given an example case, ≤ 30% of cardiologists correctly diagnosed heterozygous FH.
- Among adults with HoFH, median age at diagnosis 12.6 years.

Newborn Screening is not like testing at any other time.

Diseases that are “good candidates” for newborn screening generally cause irreversible disease or death *in childhood*.

The strongest candidates are for diseases that require treatment at time of diagnosis (ie, neonatal period).

Almost all conditions on newborn screening panels are recessive genetic diseases.

They're *rare* – vast majority of newborns have a normal result.

Relatives are carriers (one copy of a mutated gene) – generally healthy.

Informed consent?

No countries require written informed consent for newborn screening.

Most countries try to avoid controversy with their screening programs and research about newborn screening.

Please fill out the form entirely with **LEGIBLE** and **ACCURATE** information

FORM EXPIRATION DATE: 2022-05-31

Submitter Use

U452201

Baby's Name (LAST / FIRST)		Multiple births (Twin, etc.)	SEX	Baby's Birthdate	
LAST	FIRST	# ____ OF ____	F M	MM / DD / YY	Time (Military)
Baby's ID # (MRN/Ait. ID)		Baby's Primary Care Provider NPI#			
Specimen Collection Date		PCP's Clinic Name			
MM / DD / YY		City Phone # ()			
Mother's Name (LAST / FIRST)		Ordering Physician/NPI#			
LAST		FIRST		LAST FIRST / NPI	
Weight at birth (grams)	Gestational age at birth	Baby's Race		Hispanic?	
g	wks	(Circle all that apply)		N Y	
Baby in ICU?		Repeat Specimen?		Transfusion(s)? (any product)	
N Y		N Y		N Y	
Place of Birth (required)		Mother's Hep B Surface Antigen		Baby on TPN now?	
NAME		CITY / STATE		(HBsAg): Neg Pos	
Blood Not Submitted (mark one)		Pulse Ox Screen Date		Time (Military)	
☐ Declined ☐ Transferred ☐ Deceased		MM / DD / YY		Pass Fail	
☐ Other		Not Screened (mark one)		☐ Declined ☐ Transferred ☐ Deceased	
Hearing Screen Date		☐ Echo normal ☐ Confirmed heart disease ☐ Other			
Circle Hearing Screen Method		Right Ear		Hearing Not Screened (mark one)	
ABR OAE BOTH		☐ Pass ☐ Refer		☐ Transferred ☐ NICU ☐ Declined	
		Left Ear		☐ Deceased ☐ Other	
		☐ Pass ☐ Refer			
WI State Laboratory of Hygiene 465 Henry Mall Madison WI 53706					

This box for Newborn Screening Laboratory use only

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- Do not place cover over circles until blood is completely dry.
- Do not remove this cover.
- Do not touch sample area.
- Do not use if damaged.



- Dried blood spot (DBS) specimen collected after 24 hours of life
- Card is air dried and mailed to central lab for testing
- All results reported to baby's pediatrician
- Positive result activates the relevant, pre-identified specialist health care team

Newborn Screening for HoFH is currently only being done in research studies.



JAMA Cardiology | **Brief Report**

Genetic Diagnosis of Familial Hypercholesterolemia in Residual Newborn Dried Blood Spots

Amy L. Peterson, MD, MS; Vanessa Horner, PhD; Michael R. Lasarev, MS; Xiao Zhang, PhD; Stephen E. Humphries, PhD; Robert D. Steiner, MD; Megan Benoy, MGCS, CGC; Jessica Tumolo, PhD; Patrice K. Held, PhD; Xiangqiang Shao, PhD



Genetic testing for HoFH.

Cholesterol first, then genetic testing for HoFH and HeFH.

Diagnosing HoFH and HeFH at birth could mean:



- Over 99% of the population is tested for FH.
- HoFH can be diagnosed at birth with appropriate treatment started.
- The genetic nature of the disease can be made clear to families and clinicians.
- The baby's relatives could be diagnosed *decades* earlier.
- A heart-healthy lifestyle can be started at birth, and for HeFH, statin therapy at 6 years of age.

Diagnosing HoFH and HeFH at birth could mean:



- Misinformation or misunderstanding of FH, particularly if health care system not prepared.
- Difficulties with access to life insurance, or discrimination due to a preexisting condition.
- Family disruptions due to non-paternity.
- We don't know if babies with high cholesterol but **no** genetic mutation found will later have high cholesterol that looks like HeFH or HoFH.
- For HeFH, feelings of anxiety or helplessness from time of diagnosis until initiation of statin therapy.
- Concerns about individual privacy and informed consent.

Effective intervention to lower cardiovascular risk in childhood may represent the single biggest opportunity we have to positively impact human health.

