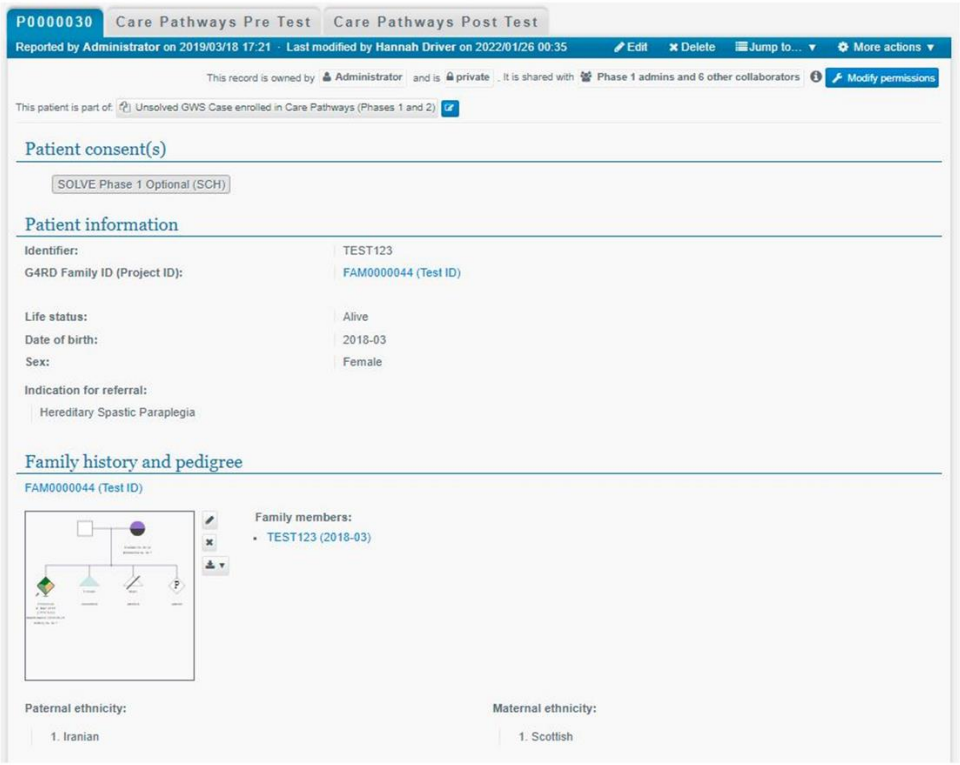
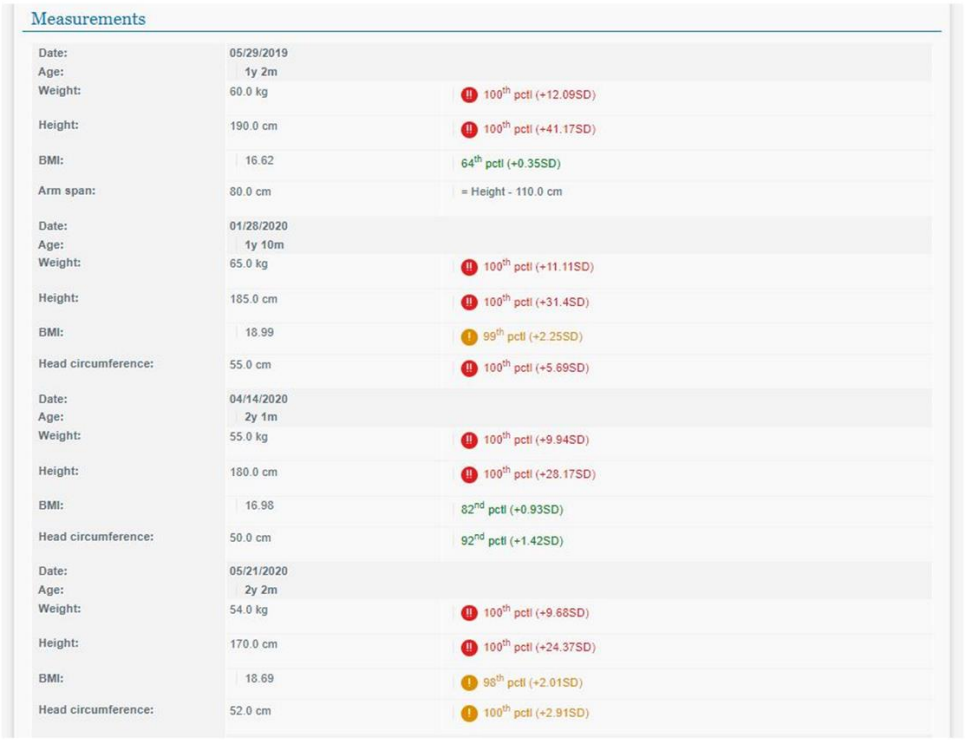
Supporting Figures

a)

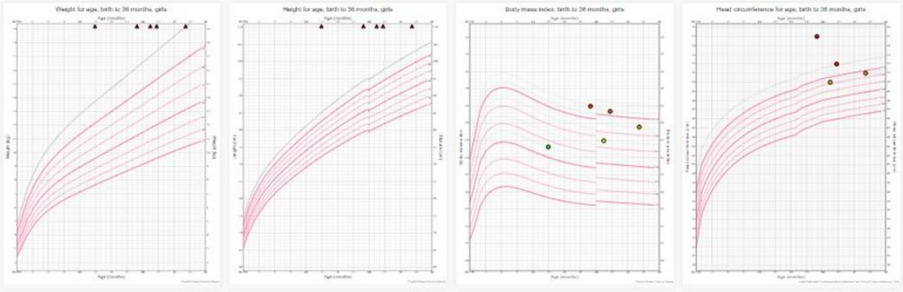


b)



c)

GROWTH CHARTS



Clinical symptoms and physical findings

GROWTH PARAMETERS

Weight for age
Increased body weight (>+2SD)
Stature for age
Tall stature (>+2SD)

EYE DEFECTS

Coloboma
★ Lens coloboma
Childhood onset

EAR DEFECTS

★ Preauricular pit

CUTANEOUS

★ Hypopigmentation of the skin
★ Generalized hypopigmentation

RESPIRATORY

Congenital diaphragmatic hernia
NO Abnormal lung morphology

BEHAVIOR, COGNITION AND DEVELOPMENT

Delayed gross motor development
Delayed speech and language development
Autism
Behavioral abnormality

d)

Genotype information

LIST OF GENES

	Gene	Status	Strategy	Comments
1	SRCAP	Candidate		
2	PSMB6	Confirmed causal		

▾SUGGESTED GENES

Click on terms below (extracted from the phenotypic description) to disable or re-enable their contribution in the gene search results.

Tall stature (194) Coloboma (300) Behavioral abnormality (2250) Autism (196) Delayed speech and language development (895) Congenital diaphragmatic hernia (102) Hypopigmentation of the skin (231) Delayed gross motor development (198) Increased body weight (529) Preauricular pit (57) Generalized hypopigmentation (50) Lens coloboma (1)

DOWNLOAD

Exclude Tested Negative And Rejected Candidate Genes

Results 1 - 10 out of 2157 per page of 10

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Matching genes	Associated phenotypes
PTEN	Autism; Behavioral abnormality; Coloboma; Delayed gross motor development; Hypopigmentation of the skin; Increased body weight; Tall stature
HERC2	Autism; Behavioral abnormality; Delayed gross motor development; Delayed speech and language development; Generalized hypopigmentation; Hypopigmentation of the skin; Increased body weight
MAGEL2	Autism; Behavioral abnormality; Delayed gross motor development; Delayed speech and language development; Generalized hypopigmentation; Hypopigmentation of the skin; Increased body weight
NIPBL	Autism; Behavioral abnormality; Coloboma; Congenital diaphragmatic hernia; Delayed speech and language development; Increased body weight
SH2B1	Autism; Behavioral abnormality; Coloboma; Congenital diaphragmatic hernia; Delayed speech and language development; Increased body weight
SIN3A	Autism; Behavioral abnormality; Coloboma; Congenital diaphragmatic hernia; Delayed speech and language development; Increased body weight
EP300	Autism; Behavioral abnormality; Coloboma; Delayed gross motor development; Delayed speech and language development; Increased body weight
ATP6V1A	Autism; Behavioral abnormality; Coloboma; Delayed gross motor development; Delayed speech and language development; Tall stature
RERE	Autism; Behavioral abnormality; Coloboma; Delayed speech and language development; Increased body weight; Tall stature
AKT1	Autism; Behavioral abnormality; Coloboma; Hypopigmentation of the skin; Increased body weight; Tall stature

Results 1 - 10 out of 2157

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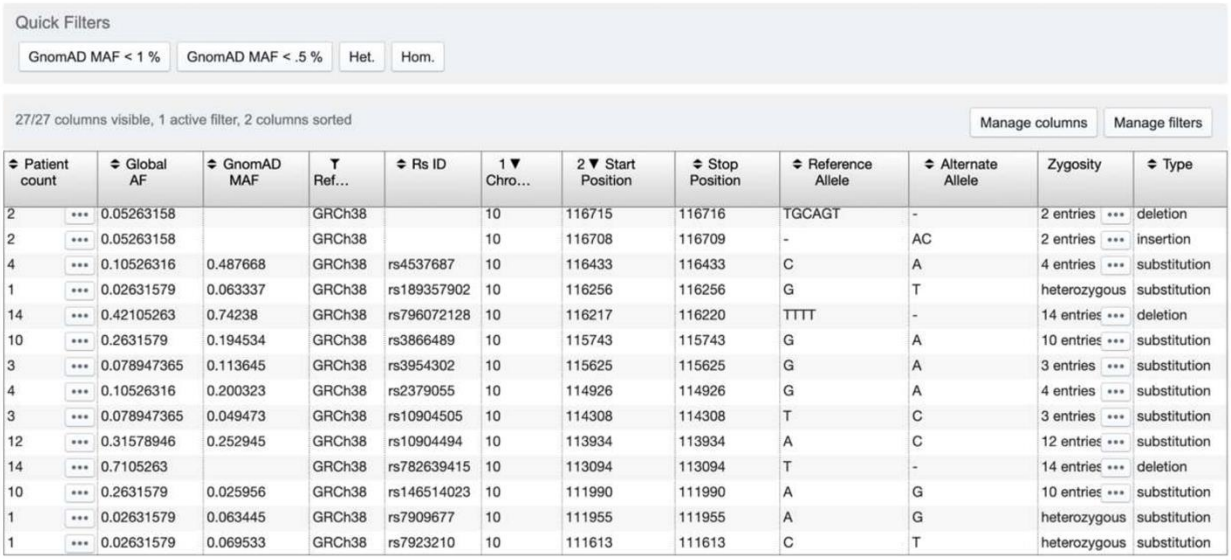
Diagnosis

Clinical diagnosis (ORDO)	1661 X-linked corneal dermoid
Final diagnosis (OMIM)	425500 HAIRY EARS, Y-LINKED

Supp. Figure S1a-d. Genomics4RD records. Genomics4RD records can store and display a breadth of participant information, including, but not limited to: a) participant consent types, coded participant demographic information, family history and pedigrees; b) clinical measurements; c) growth charts, clinical symptoms and physical findings (using HPO terms, starred for relevance); and d) genotype information, suggested candidate genes based on HPO-gene associations, and diagnoses. The Care Pathway modules are available as separate forms, accessible at the top of participant records enrolled in the study (see a).



Patient count	Global AF	GnomAD MAF	Ref...	Rs ID	Chro...	Start Position	Stop Position	Reference Allele	Alternate Allele	Zygosity	Type
2	0.05263158		GRCh38		10	116715	116716	TGCAGT	-	2 entries	deletion
2	0.05263158		GRCh38		10	116708	116709	-	AC	2 entries	insertion
4	0.10526316	0.487668	GRCh38	rs4537687	10	116433	116433	C	A	4 entries	substitution
1	0.02631579	0.063337	GRCh38	rs189357902	10	116256	116256	G	T	heterozygous	substitution
14	0.42105263	0.74238	GRCh38	rs796072128	10	116217	116220	TTTT	-	14 entries	deletion
10	0.2631579	0.194534	GRCh38	rs3866489	10	115743	115743	G	A	10 entries	substitution
3	0.078947365	0.113645	GRCh38	rs3954302	10	115625	115625	G	A	3 entries	substitution
4	0.10526316	0.200323	GRCh38	rs2379055	10	114926	114926	G	A	4 entries	substitution
3	0.078947365	0.049473	GRCh38	rs10904505	10	114308	114308	T	C	3 entries	substitution
12	0.31578946	0.252945	GRCh38	rs10904494	10	113934	113934	A	C	12 entries	substitution
14	0.7105263		GRCh38	rs782639415	10	113094	113094	T	-	14 entries	deletion
10	0.2631579	0.025956	GRCh38	rs146514023	10	111990	111990	A	G	10 entries	substitution
1	0.02631579	0.063445	GRCh38	rs7909677	10	111955	111955	A	G	heterozygous	substitution
1	0.02631579	0.069533	GRCh38	rs7923210	10	111613	111613	C	T	heterozygous	substitution

Supp. Figure S2. Genomics4RD variant store. The variant store aggregates variants across participants in Genomics4RD. Data depositors can multi-filter the store based on variant annotations, zygosity, and counts across participants.