Supplemental Table 2. Epidemiology of GSDs			
Condition	**Overall frequency**	**Frequency in specific ethnicities**	**Variants common in specific ethnicities**
GSD 0	Unknown	Unknown	Unknown
GSD I	Incidence ~1 in 100,000 (80% GSD Ia; 20% GSD Ib)[89,90]	Ashkenazi Jewish: Prevalence 1 in 20,000[90]	1) *G6PC1*; c.79delC (p.Gln27ArgfsTer9). 1 of 3 variants that comprise 21% of pathogenic variants in individuals of European ancestry.[90]
			2) *G6PC1;* c.247C>T (p.Arg83Cys). 32% of variants in European population[90,91] and 96% of variants in Jewish population (carrier frequency 1.4% in Ashkenazi Jewish population[92])
			3) *G6PC1;* c.248G>A (p.Arg83His). 38% of variants in Chinese population.[90]
			4) *G6PC1*; c.379_380dupTA (p.Tyr128ThrfsTer3). 50% of variants in Hispanic population.[90]
			5) *G6PC1*; c.562G>C (p.Gly188Arg). 1 of 3 variants that comprise 21% of pathogenic variants in individuals of European ancestry.[90]
			6) *G6PC1*; c.648G>T (p.Leu216Leu). Approximately 85-88% of variants in Japanese population and 36-40% of variants in Chinese population.[90,93-95]
			7) *G6PC1*; c.1039C>T (p.Gln347Ter). 1 of 3 variants that comprise 21% of pathogenic variants in individuals of European ancestry.[90,93]
			8) *SLC37A4*; c.352T>C (p.Trp118Arg). 37-50% of variants in Japanese population.[90,93,96]
			9) *SLC37A4*; c. 1015G>T (p.Gly339Cys). ~15-21% of variants in mixed European population and 29% in German population.[90,93]
			10) *SLC37A4*; c.1042_1043delCT (p.Leu348Valfs*53). ~27-31% of variants in mixed European population and 32% of variants in German population.[90,93]
GSD II (Pompe disease)	~1 in 40,000[16,97,98] historically (combined IOPD and LOPD), but the epidemiology of Pompe disease has been informed by NBS.[99] A population genetics approach estimated predicted genetic prevalence 1 in 23,232 overall.[99]	Infantile form appears to have higher incidence in African-American and Chinese ancestry.[16]	1) *GAA*; c.-32-13T>G. ~60-90% of alleles in individuals with LOPD (the variant is more common in individuals of European ancestry).[100-104]

		LOPD appears to be more common in The Netherlands.[16,97,105]	2) *GAA*; c.525delT (p.Glu176ArgfsTer45). ~5% of pathogenic alleles in the US population[106]
		Population genetics study estimated prevalence of overall Pompe disease stratified by ethnicity.[99]	3) *GAA*; c.1935C>A (p.Asp645Glu). ~80% frequency of variant in Chinese patients with IOPD.[107,108]
			4) *GAA*; c.2482_2646del (p.Gly828_Asn882del). ~26% of affected Europeans (~13% of alleles).[109]
			5) *GAA*; c.2560C>T (p.Arg854Ter). Common variant among individuals of African ancestry (in one study, ~47% of alleles)[110]
GSD III	Prevalence 1 in 100,000 (85% GSD IIIa; 15% GSD IIIb)[32]	Nunavik Inuit prevalence: ~1 in 2,500[111]	1) *AGL*; c.2590C>T (p.Arg864Ter). Approximately 10.3% of pathogenic variants in the US.[112,113]
		Faroe Islands prevalence: ~1 in 3,100[114]	2) *AGL*; c.4260-12A>G. Approximately 5.5% of pathogenic variants in the US.[113,115]
		Prevalence among individuals of North African Jewish ancestry in Israel: 1 in 5,400[27,116]	3) *AGL*; c.3965delT (p.Val1322AlafsTer27). Approximately 6.7% of pathogenic variants in the US.[115]
			4) *AGL*; c.3682C>T (p.Arg1228Ter). Approximately 5.2% of pathogenic variants in the US.[27]
			5) *AGL*; c.1222C>T (p.Arg408Ter). Founder variant in the Faroe Islands.[114]
			6) *AGL*; c.4456delT (p.Ser1486ProfsTer18). Founder variant in individuals of Inuit ancestry[111] and North African Jewish ancestry.[116]
			7) *AGL*; c.2309-1G>A. 11.8% of alleles in individuals of European ancestry.[113]
			8)*AGL*; c.1384delG (p.Val462Ter). ~50% of alleles in individuals of Hispanic ancestry (2 of 4 individuals homozygous).[113]
GSD IV	~1 in 600,000 to 1 in 800,000.[33,35]	APBD prevalence is thought to be more common among individuals of Ashkenazi Jewish ancestry.[117]	1) *GBE1*; c.986A>C (p.Tyr329Ser). APBD founder variant in the Ashkenazi Jewish population.[117] The estimated carrier frequency is 1 in 48 among individuals of Ashkenazi Jewish ancestry.[33]
			2) *GBE1*; c.2053-5289_2053-5297delins TGTTTTTTACATGACAGGT. APBD founder variant in the Ashkenazi Jewish population.[117]

GSD V	Prevalence estimated to be 1 in 100,000 in Dallas/Fort Worth of USA.[37] Prevalence in Spain estimated ~139,543.[118]	Unknown	1) *PYGM*; c.148C>T (p.Arg50Ter). The most frequent variant seen in individuals of European ancestry. 31% of variants in Netherlands, 43% of variants in Italy, 55% of variants in Spain, 58% of variants in German, 60-63% of variants in US, 68-72% of variants in France, 77-81% of variants in UK.[119^]
			2) *PYGM*; c.613G>A (p.Gly205Ser). Varying reports of frequency in different populations.[119] 9% of variants in Spain.[118]
			3) *PYGM*; c.2392T>C (p.Trp798Arg). Seen in 10% of Spanish individuals with GSD V.[118]
			4) *PYGM*; c.2128_2130delTTC (p.Phe710del). 68% of variants in individuals of Japanese ancestry.[120]
			5)*PYGM*; c.1A>G (p.Met1Val). In one study of 67 patients from Turkey, this was the most common variant identified (27 patients from 11 families).[121]
GSD VI	Best prevalence estimate appears to be 1 in 100,000[40] but estimates vary.	Prevalence ~1 in 1,000 in Mennonite population.[43,122]	1) *PYGL*; c.1620+1G>A. Founder variant in Mennonite population.
GSD VII	Rare	Appears to be more common in individuals of Ashkenazi Jewish ancestry.[36]	2 common variants in affected individuals of Ashkenazi Jewish ancestry. One variant, affecting exon 5 splicing, comprises about 68% of pathogenic variants in this population.
GSD IX	Liver PhK deficiency has had estimated frequency of 1 in 100,000.[40,49] Muscle PhK deficiency appears rare.[49] *PHKA2* accounts for ~75% of hepatic GSD IX cases.[40,49] *PHKA2* is X-linked, and disease is more common in males.	Unknown	1) *PHKA2*; c.3614C>T (p.Pro1205Leu). Common variant in Dutch patients.[123]
GSD X	Rare	While most individuals reported have been of African American ancestry, affected individuals of other ethnicities have been described.[124]	Unknown. Although most affected African American individuals had the p.W78X variant, GSD X has been described in very few reports.
GSD XI (LDHA deficiency; note that FBS is sometimes called GSD XI also)	Rare	Unknown. One study of 3,776 individuals in Japan suggested a carrier frequency of 0.185%.[125]	Unknown. A 20 base pair deletion in exon 6 has been identified in multiple Japanese families.[126]
GSD XII	Rare	Unknown	Unknown
GSD XIII	Rare	Unknown	Unknown
GSD XV	Rare	Unknown	Too few individuals have been reported to confidently comment on the presence of a founder variant. In a paper describing 7

			unrelated individuals of different ancestries, 4 had the c.143+3G>C variant (2 homozygous and 2 compound heterozygous with c.143+3G>C on one allele).[73] 2 sisters of Italian ancestry whose parents were first cousins were homozygous for c.143+3G>C.[127] 5 patients (4 families) of Sardinian ancestry were reported as homozygous for c.143+3G>C.[74] It is possible c.143+3G>C could be a founder variant in individuals of Italian ancestry or other ancestry, but future reports may be informative.
FBS	Rare	Unknown	1) *SLC2A2*; c.157C>T (p.Arg53Ter) was reported to be homozygous in 6 unrelated patients in Sudan (the study included 11 patients from 10 families). It was suggested this may be a Sudanese founder variant.[128]

Supplemental Table 2. Different publications report different terms for epidemiology, such as prevalence and incidence, which have notable distinctions. We have tried to either use the nomenclature of the cited studies or a broad term. ^Numerous studies evaluating the frequency of the *PYGM* variant p.Arg50Ter in different countries are reviewed in the Nogales-Gadea et al. (2015) publication cited. GSD, glycogen storage disease; LOPD, late-onset Pompe disease; IOPD, infantile-onset Pompe disease; APBD, adult polyglucosan body disease; PhK, phosphorylase kinase; LDHA, lactate dehydrogenase subunit A; FBS, Fanconi–Bickel syndrome.