



The ASF Alport Patient Registry, which launched August 24, 2023, is open to individuals living with Alport syndrome in the U.S. (+ territories and outlying islands), at all ages and stages of disease.

Patient participation helps attract and advance Alport syndrome research and clinical trials. To get prepared for the registry, ASF has compiled some quick notes and tips to help simplify the process of inputting your data.

General Notes

- The ASF Alport Patient Registry is open to individuals with a confirmed clinical diagnosis of Alport syndrome. This can include a positive diagnosis via genetic testing and/or biopsy, and/or a diagnosis from a medical professional based on an individual's symptoms and/or family history.
- Security of patient data is a top priority. All data is deidentified, and no electronic health records or social security numbers are requested or connected. Participants' home address is optional.
- All registry enrollees are strongly encouraged to add "asfalportpatientregistry@healthie.net" to their email contacts to avoid missing important communications about registry participation.
- Patients are encouraged to use a desktop or laptop with an updated browser when accessing the ASF Alport Patient Registry, as opposed to a tablet or mobile phone/device.
- If you need to change your password for any reason at any time, simply select the "Did you forget your password?" prompt [on the registry login screen](#). Follow the on-screen instructions to create a new password.

- If you need to change the login email associated with your registry profile, please email info@alportsyndrome.org for further assistance. All other technical inquiries can also be directed to this email address.
- Parents can also enroll their child/children under the age of 18 with Alport syndrome *at any time* by clicking their full name at the top of the registry Dashboard (main page) and then selecting “Add Participant.”
- Note to parents/caregivers: If you have enrolled a child (or children) in our registry since March 2022 and they have reached 18 years of age since then, please encourage them to [enroll as a new member of the patient registry using this link](#). A unique email address (not associated with the user's parent/caregiver account) will be required for the adult child to enroll.
- It's important to remember the ASF Alport Patient Registry will continually capture a registrant's health data over time via open review periods spaced approximately one hundred eighty days apart. Around six months after enrollment in the registry, you will receive an email from “asfalportpatientregistry@healthie.net” alerting you to re-review the survey questions and update them with answers pertaining to various aspects of your health data. You will not need to re-enter all data (data fields will be pre-populated with your previous answers). Simply review and update, as needed. For example, updating your latest kidney lab values (eGFR, creatinine, etc.) and medication types and doses is critical to Alport research. Once you review/update your data and hit “Finish Now,” your answers will remain locked until the next open review period.
- Genetic testing results can be uploaded *at any time* (in PDF, JPEG, or PNG format) by visiting “Profile” → “Upload My Files. Note: If your genetic testing results were inconclusive/negative but you were still clinically diagnosed by a medical professional as having Alport syndrome, it is important for you to upload your genetic testing file(s). Research has shown “intronic” Alport syndrome variants may not be detected via whole-exome sequencing. Collecting data on intronic genetic variants is critical to advancing our understanding of Alport syndrome genetics.
- Audiograms can also be uploaded *at any time* (in PDF, JPEG, or PNG format) by visiting “Profile” → “Upload My Files.” Multiple files can be uploaded at once (ex. numerous historical audiogram results) to provide more robust data. Note: If you do not have hearing loss, but are being monitored due to Alport syndrome and have had a hearing test in the past, it is also valuable to provide your latest audiogram findings to show a baseline of no hearing loss. Capturing no hearing loss and/or onset of hearing loss are both extremely valuable.
- Mandatory questions in the registry are indicated by a red asterisk (*). Even if you choose not to answer the optional questions, your registration as an Alport

patient is still very valuable and offers you the opportunity to opt in or opt out of being notified about specific clinical trials for which you/your child(ren) may be qualified.

Tips for Quick Data Entry

Having the following information on hand will help you quickly move through the registry survey questions:

- Your current stage of CKD (chronic kidney disease). The registry platform will have a reference list of estimated glomerular filtration (eGFR) rates and which CKD stage they correspond to.
- Roughly how many years ago a doctor or health care provider told you that you have Alport syndrome.
- Your family history of Alport syndrome. If you are unaware, it will be helpful to find out if other family members have experienced symptoms and/or been diagnosed with Alport syndrome. Examples include: Mother, father, sister, aunt, grandfather, cousin, uncle, etc.
- Any history of misdiagnosis.
- The names and dosages of current Alport treatment medications. Prior treatment history can also be logged.
- If you or anyone in your family has experienced an aortic aneurysm.
- Date of initiation of dialysis and/or date of kidney transplant(s), if applicable.
- At what age you became aware of your hearing loss (age range estimate), if applicable.
- The type of Alport syndrome that appears on your genetic testing results, if applicable.
- If applicable, personal history of pregnancy complications.
- If you are able to share any diagnosed eye abnormality (ex. anterior lenticonus, macular degeneration or macular holes, dot and fleck retinopathy, temporal retinal thinning).

Laboratory Results You Will Be Asked to Share

You will be asked to input your lab results and units of measure exactly as they appear on your report. You may need to access your clinic's online health portal to see units of measure associated with lab values, as paper printouts may omit units for brevity. You will also be required to enter the year and month of your latest lab values, with the option to add the specific date.

- Most recent eGFR (estimate glomerular filtration rate). You can choose an estimated range or option to type in exact value, if known. The standard unit of eGFR measurement is milliliter/minute/1.73 meter².
- Most recent blood creatinine value and unit of measurement. Units of measure are mmol/L, μ mol/L, mg/dL, mg/L, and μ g/mL.
- Most recent blood potassium value and unit of measurement (mmol/L or mEq/L).
- Most recent hemoglobin value and unit of measurement (g/L or g/dL).
- Most recent urine albumin-creatinine ratio (UACR) value and unit of measurement (mg/g or mg/mmol).